Indiana State Department of Health
Maternal and Child Health Services
The State Department on Genetic Disease Program

Planning for Genetic Services in Indiana

Evaluation Report, September 2001
Revision October 9, 2001

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APPENDICES

1.0 INTRODUCTION: Planning for Genetic Services In Indiana

Rapid advances in technology and discoveries fueled by the Human Genome project present unique challenges to health policy leaders and decision makers in the state. These new technical advances are changing the delivery of health care and the economics of health care provision, and they have broad social implications. Improved birth defect surveillance programs, advancing knowledge about the genetic contribution to adult onset chronic diseases, and an increasing understanding of the interaction of the environment with genetic predispositions and behavioral risk activities require leaders' attention to the problems associated with access problems and changing levels of service needs.

In 1997, the Centers for Disease Control and Prevention (CDC) developed the Office of Genetics and Disease Prevention to spotlight the emerging role of genetics in the practice of public health in the United States and to provide internal coordination and promote external partnerships in those activities that are related to genetics and disease prevention and health promotion. This action was recommended in an agency-wide strategic plan that outlines a conceptual framework for a public health program in genetics (CDC, 1997). The strategic plan is based on the assumption that virtually every human disease of important public health impact is the result of the interaction between human genetic variation and the environment. It is also based on the assumption that the use of genetic information in public health is feasible and worthwhile in diagnosing, treating and preventing disease, disability, and death among people who inherit specific genotypes. Prevention includes the utilization of medical, behavioral, and environmental interventions in order to reduce the risk for disease among those individuals who are susceptible because of their genetic makeup. The plan supports the accountability of genetic tests and services, including appropriate family history assessment and genetic counseling, for promoting health and preventing disease in diverse communities. The plan assumes that much of the delivery of genetic tests and services will be performed within the context of the changing health care system, including managed care organizations, rather than under the auspices of public health agencies. The role of public health organizations will emphasize assessing the health needs of populations, assuring the quality of genetic tests and services and evaluating the impact of interventions.

To address the need to integrate and meld the genetic research advances with the public health agencies' concerns for health intervention, the Indiana State Department of Health (ISDH) was awarded federal funds through the Maternal and Child Health Bureau (MCHB) (Genetics Planning Grant-CFDA # 93.110A). This project supported the development of an information and data infrastructure to guide and assist public health professionals with the identification of infants and children (0-3 years of age) with birth defects. The project expanded its focus beyond initial concerns with mortality and morbidity issues to a broader challenge of developing a Genetics Advisory Committee, conducting a needs assessment, establishing new data bases and linkages, and finally developing an Indiana State Genetics Plan.

Current statistics regarding genetic disorders in the nation and in the state of Indiana raise important policy concerns. Each year in the United States, about 150,000 birth defects occur resulting in approximately 6,200 deaths. Birth defects are the leading cause of infant mortality, accounting for more than one in five infant deaths. The United States' infant mortality ranks

worst among the G-7 industrialized nations and 25th in the world (NCHS, 1999). Cost estimates for the families and the nation range up to eight billion dollars annually (Pew Environmental Health Commission, na; Waitzman, Scheffler & Romano, 1996). In 1997 in Indiana 83,436 births were recorded with an estimated 3,337 birth defects and 675 infant deaths (March of Dimes, na). For those individuals who survive, service systems must respond to service needs for survival and quality of life services. Indiana's First Step Program which provides early intervention for families who have infants and toddlers with developmental delays or who are developmentally vulnerable identify 255,572 children ages birth to three in their target population. Of that total number, 35,582 are eligible for services but only 14,626 are enrolled in the program. The average days from referral to enrollment is 42.9 days. Although not all developmental delays are the result of birth defects, a significant number of these children could be helped with better birth defects surveillance programs and services.

The effects of genetic disorders can be far-reaching and sometimes devastating to the families. The existence of a genetic condition indicates an increased risk for other family members and in future pregnancies. Individuals affected usually have long-term health, social service and education needs, resulting in out-of-pocket expenses for families, third-party payers and/or public funds.

Understanding and assessing the needs of genetics services in the state of Indiana would include: 1) evaluating that population's genetic service needs, 2) determining whether or not gaps in the delivery of genetics services exist, 3) determining where any known gaps may exist, 4) assessing gaps in provider and consumer knowledge base, and 5) making recommendations on how to reduce or eliminate those indicated needs. To accomplish the assessment goal, elements of three traditional needs assessment strategies were used. The discrepancy model approach or gap model, identified levels of need and examined performance measures of the current system to identify gaps. The marketing model approach utilized community input from various sources to identify perceptions and preferences. And, finally, elements of a decision-making model that identified problems and provided a group decision making process to rank, prioritize and develop a plan of action based on the information developed for this report will culminate in a state-wide plan (Fineman & Doyle, 2000). A wealth of information was drawn from primary and secondary sources, direct interviews, and surveys to identify the level of need, types of service provision, perceptions of gaps and concerns about services and need for training.

This report details those activities. It is organized into five sections. Following this introduction, Section 2.0 describes the prevalence of various health problems that are associated with genetic deficiencies and attempts to identify the burden of disease and disability in different communities. National data sets are reviewed along with various state information systems. Section 3.0 describes and graphically illustrates what types of services are offered and where they are located. Section 4.0 reports three primary data gathering activities that tap into community perceptions, community leaders' concerns, and provider practices. The final section 5.0 contains recommendations for action and a plan for the conduct of the Genetics Advisory Committee activities for the coming months. In addition, a legislative benchmarking activity

was performed to compare Indiana's policy initiatives with other states in the nation. The results of that analysis are contained in Appendix A.

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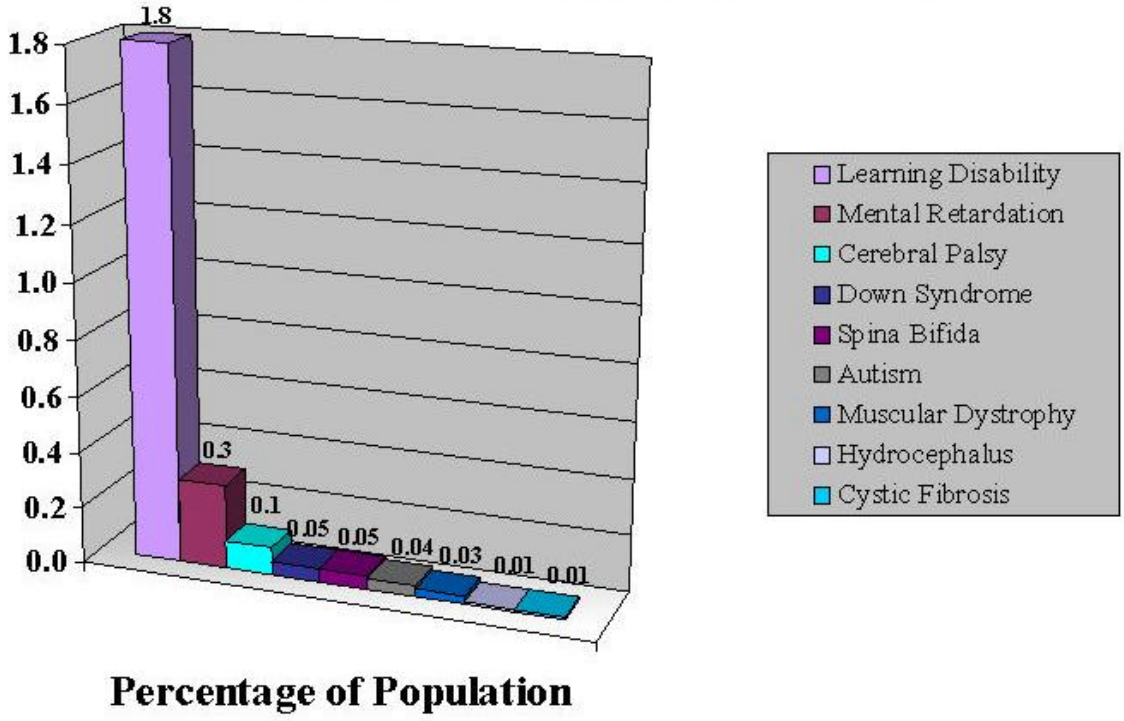
2.0 INCIDENCE & PREVALENCE PATTERNS

Challenges abound in identifying accurate incidence and prevalence rates for various birth defects and diseases with genetic foundation. Frequency of birth defects is noted to be three to five percent or one out of twenty newborns or infants, and cancer-related conditions resulting from genetic abnormalities are one out three. Some defects or genetic conditions are extremely low in incidence among the general population. Sample sizes necessary to obtain any cases of the (statistically) rarely occurring conditions are prohibitive in cost and in time for data collection. The technical staff utilized existing national data bases for the most general of prevalence rates and state specific data systems that identify individuals with birth defects/ genetic conditions. National data sets cannot be accurately generalized to a state/local level without synthetic estimation techniques. State and local data bases may suffer from reporting inconsistencies and constitute a probable undercount due to reliance only on officially reported cases. Nonetheless, even with these caveats, these sources represent some of the best available information for planners and decision makers.

An initial examination of prevalence rates for typical birth defects/genetic conditions and birth defects in the general population is provided by the National Study of Health and Disability, 1995. Figure 2.1 illustrates the extremely low percentage of individuals in the nation who report these problems. The prevalence rates range from a high of 1.8 percent (1.8%) for learning disability to a low of 0.01 percent (0.01%) for hydrocephalus and cystic fibrosis. Other difficulties identified in national studies are graphically summarized in Appendix B.

Figure 2.1: National Study of Health & Disability
Prevalence in Population - All Ages



2.1 Prevalence Studies in Indiana

The prevalence of genetic conditions is assessed in several different ways in Indiana, some of which may not be adequate. Congenital anomalies (structural birth defects) are identified primarily by data recorded on birth certificates. Screening programs exist to identify selected metabolic disorders, hearing problems, and prenatal exposure to controlled substances. Developmental disorders are not specifically screened, but this chapter will use data on program participation (in the "First Steps" program of Family and Social Service Administration--FSSA) to establish a lower-bound estimate of prevalence. Prevalence of congenital infections is not currently documented. Table 2.1 gives incidence estimates by the March of Dimes for most categories of birth defects.

Table 2.1: March of Dimes Estimates of Birth Defects for 2000

	Estimated Incidence per 10,000 births			
tructural/Metabolic				
Spina bifida	5.0	or	1 in 2,000	births
Anencephaly	1.3	or	1 in 8,000	births
Nervous system and eye	42.6	or	1 in 235	births
Respiratory tract	11.1	or	1 in 900	births
Heart and circulation	87.0	or	1 in 115	births
Genital and urinary tract	74.1	or	1 in 135	births
Cleft lip/palate	10.8	or	1 in 930	births
Club foot	13.6	or	1 in 735	births
Muscles and skeleton	76.9	or	1 in 130	births
Chromosomal syndromes	16.7	or	1 in 600	births
Down syndrome (Trisomy 21)	11.1	or	1 in 900	births
Metabolic disorders	2.9	or	1 in 3,500	births
Phenylketonuria (PKU)	0.8	or	1 in 12,000	births
ongenital Infections				
Congenital syphilis	5.0	or	1 in 2,000	births
Congenital HIV infection	3.7	or	1 in 2,700	births
Congenital rubella syndrome	0.1	or	1 in 100,000	births
ther				
Rh disease	7.1	or	1 in 1,400	births
Fetal alcohol syndrome	10.0	or	1 in 1,000	births

Note: all numbers are based on the best available estimates, which underestimate the incidence of many birth defects. Sources: March of Dimes Perinatal Data Center, 2000. This data is from an unpublished review of the literature and information from various state and regional birth defects surveillance systems (California, Iowa, Metropolitan Atlanta, New York, and Texas).

2.2 Congenital Anomalies (Structural Birth Defects)

It has been shown that the exclusive use of birth certificate data is inadequate to document incidence prevalence of congenital anomalies (Watkins et al, 1996). Experience in Indiana has also shown that birth certificate information on congenital anomalies does not agree well with information on the medical record (Zollinger, 2000). However, birth certificate data are still the only means used to identify the incidence of congenital anomalies in Indiana. In this study the Indiana system is compared to systems in other states.

A directory of birth defects surveillance programs compiled by the State Services Branch of the Division of Birth Defects and Pediatric Genetics was published in the January / February 2000 issue of *Teratology*. In the same issue, The National Birth Defect Prevention Network (NBDPN) presented a compilation of birth defects surveillance data from selected states 1989-1996. By considering both the characteristics of the surveillance programs and the availability of data, four states were chosen for a comparison study of congenital anomalies. In this section of the report, Illinois, Iowa, Wisconsin, and Georgia are compared to Indiana. The Georgia program was selected because it is one of the oldest and most extensive surveillance systems in the county. It is an active surveillance system using data from many different sources including hospital medical records, hospital department logs, hospital discharge index, and birth certificates. The Iowa system is also an active system based on a similarly broad base of data sources. Unlike the Georgia system, which covers only births to residents of the Atlanta metropolitan area, the Iowa system is statewide. Illinois uses a combination of active and passive ascertainment. Cases are identified and reported by hospital staff, but state health department abstractors go to hospitals to abstract birth information. The Wisconsin system, like Indiana's, is passive. But unlike Indiana, Wisconsin also uses hospitalization and Medicaid claims files to document birth defects.

Figures 2.2 through 2.8, presented at the end of this section, show that Indiana has a lower reported incidence of most categories of congenital anomalies. It seems reasonable to assume that these lower incidence rates are partly due to our less intensive surveillance program. For example, Figure 2.2 shows that Indiana had the lowest rate of chromosomal congenital anomalies at 8.15 per 10,000 live births over the 1989 to 1995 period. The next-lowest rate among the comparison states was in Illinois at 12.05 per 10,000. The comparable March of Dimes estimate from Table 2.1 was 16.7. Of the seven classes of congenital anomalies presented in Figures 2.2-2.8, Indiana had the lowest rate four times and was second lowest once. In the case of Genital/Urinary anomalies (Figure 2.7), Indiana ranked third among the five states with 23.37 cases per 10,000 live births in the 1989-95 period. The two states with the most comprehensive surveillance systems, Georgia and Iowa, reported much higher rates (53.28 and 44.06 respectively).

Only in one class of anomalies did Indiana report a relatively high rate. There were 35.47 cases of Musculoskeletal/Integumental anomalies per 10,000 births in Indiana while the other states had rates ranging from 5.42 to 32.41 (Figure 2.8). One explanation for the high rate in Indiana may be the way this class of anomalies was recorded. The Indiana birth certificate allows a choice among 26 specific congenital anomalies, and eight of them are in this one category. The reporting of birth defects could be sensitive to the classification system used. The system used on the Indiana birth certificate differed substantially from the common system used in the

NBDPN compilation for the other states. In this class of anomalies, it was not clear that the NBDPN system provided complete coverage. The eight Indiana anomalies included a category for "other" anomalies in this class while the NBDPN system did not. The March of Dimes estimate for birth defects of "muscles and skeleton" was 76.9 per 10,000, much higher than Indiana's rate of 35.47. Of course, the March of Dimes estimate was for 2000 while the other estimates were for 1989-1995.

The reporting of congenital anomalies is clearly imperfect. There is also no good way to distinguish real differences in rates from differences in the comprehensiveness of the surveillance programs. Figures 2.2 through 2.8 show that in most cases the rates are positively correlated with the comprehensiveness of the systems.

Comparing specific individual anomalies should be clearer than comparing classes of anomalies that may not have exactly the same definitions. Figures 2.9 through 2.13 show that Indiana had the lowest rate among the five systems for all of these anomalies in 1989-95. These figures also show how Indiana's rates have changed from 1997 to 1999 although rates for the other states are not available. In the case of anencephalus (Figure 2.9), the consistent downward trend in Indiana's rate may be real. The March of Dimes 2000 estimate from Table 2.1 was 1.3 per 10,000, approximately the same as the 1999 Indiana rate.

Although the compilation of adequate prevalence rates for Indiana is clearly problematic, it may still be interesting to see how rates vary within the state. Nine birth defects were chosen to analyze further: anencephaly, spina bifida, cleft lip, cleft palate, anal atresia/stenosis, heart defects, omphalocelle, other genital/urinary anomalies, and other nervous system anomalies (Van Meter, na). These birth defects were chosen because most have been shown to be multifactorial defects, meaning that environmental factors can play a role in the cause of the defect. The goal is to identify areas of Indiana that are at an increased risk for having a child with a particular defect and to attempt to identify potential causes of the birth defect. By having more information about potential causes and areas that are at an increased risk, the Indiana State Department of Health can target these areas for prevention. Microcephaly is another anomaly that can result from environmental factors; however, the numbers were too small to analyze.

Figure 2.14 shows the rates per 10,000 of all nine congenital anomalies over the 1994 to 1998 period. The highest rates are clearly *not* in the most urbanized areas. But there is no other obvious pattern to the zip codes with the highest rates. Since these rates are so low (around 100 to 300 per 10,000 or 1% to 3%) a zip code would have to have a fairly large number of cases before statistically significant differences from the statewide proportion could be observed even if the absolute difference in rate was quite large. Further research should aggregate data over a longer time period and focus on whether there are any attributes common to the high-rate areas.

Congenital anomalies are a leading cause of hospitalizations among children. Other states' surveillance systems focus on hospital data to supplement data on the birth certificate. Figures 2.15 and 2.16 give some idea of the extent and distribution of hospitalizations for congenital anomalies in Indiana. Figure 2.15 shows the proportion of the 4,082 such hospitalizations in 1998 and 1999 in each of the seven categories reported in Figures 2.2 through 2.8. Circulatory and respiratory anomalies lead to the most hospitalizations. It should be noted that each

hospitalization is classified by its primary ICD9CM diagnosis code. These are only the congenital anomaly codes (ICD9CM 740 to 759). It is very possible that a child with a congenital anomaly could be hospitalized under some other primary diagnosis or that the primary congenital hospitalization diagnosis (say a congenital heart condition) might not be the child's primary anomaly (say Down Syndrome). Figure 2.16 shows the distribution of these same hospitalizations within the state. Fully forty-four percent (44%) were at Riley Hospital for Children in Indianapolis. Another twenty-four percent (24%) were at other hospitals in the greater Indianapolis area. This concentration of hospitalizations would make it easier to implement a hospital-based surveillance system. It also shows that hospital services may be poorly distributed to serve people outside the Indianapolis area.

2.3 Metabolic Birth Defects

Table 2.2: Metabolic Disorders per Genotype in Indiana 1995-1999

	Cases 1995-99	% of total	Rate per 10,000 births
Galactosemia - DD	2	0.41%	
Galactosemia - DG	124	25.25%	
Galactosemia - GG	9	1.83%	
Galactosemia - LAD	1	0.20%	
Galactosemia - UNKNOWN	35	7.13%	
Galactosemia -DG	1	0.20%	
	172	**35.02%**	**6.85**
Hyperphenylalaninemia	2	0.41%	
Hypothyroidism	155	31.57%	
Hypothroidism - RHC	4	0.81%	
	159	**32.38%**	**3.78**
Maple Syrup Urine Disease	1	0.20%	0.24
Normal Hypothyroidism	2	0.41%	0.48
Phenylketonuria	34	6.92%	
Phenylketonuria – RHC	1	0.20%	
	35	**7.12%**	**0.83**
Total births 1995-99:	**420,514**		

Indiana has conducted screening for metabolic conditions since 1965. A "Genetics Confirmed Positive Registry" (GCPR) has been implemented to document service delivery to these children. Over the 1995 to 1999 period, 491 children were born who were entered into the GCPR. Thirty-five of them were diagnosed with phenylketonuria (PKU) making the rate of PKU in Indiana 0.83 per 10,000. This compares with the rate of 0.8 found by the March of Dimes (see Table 2.1). Other metabolic disorders, primarily galactosemia and hypothyroidism, had a rate of 10.84 per 10,000 in Indiana. The full listing of metabolic disorders is shown in Table 2.2.

2.4 Screening for Hearing and Exposure to Controlled Substances

Data for the second half of 2000 were available for the hearing screening programs. About ninety-seven percent (97%) of babies born in Indiana over this period were documented as being screened for hearing loss. Of those screened, 12.7 percent (4,268 children) did not pass either an initial or a follow-up screen. Of those not passing the screening, 19.2 percent (19.2%) were referred for services. The National Health Interview Survey reports a 1.26 percent (1.26%) rate of hearing impairment in persons less than 18 years old.

2.4.1 Prenatal Exposure to Controlled Substances: Meconium Screening

Much of the material in this section is taken from the *Meconium Screening Report* prepared by ISDH pursuant to Indiana legislation PL 260-1997 and PL 273-1999 to report on results for the 2000 calendar year. The 1997 legislation established the following criteria for meconium screening:

* The infant's weight is less than 2,500 grams;
* The infant's head is smaller than the third percentile for the infant's gestational age; and
* There is no medical explanation for the above conditions.

In 1999, new legislation authorized the ISDH to establish new (additional) criteria for expanded mandatory (funded) meconium screening and required the preparation of reports on the results of the screening program. During the second half of 2000 (beginning in July) screening was also required and funded if any one of the following was present:

* Maternal current or past drug use
* No or inconsistent prenatal care
* Infant shows signs/symptoms suggestive of drug effects/withdrawal
* Unexplained abruptio placentae

More recently, the screening requirements have again been changed to include infants where any two of the above are present. Voluntary meconium screening is also provided, but it is not funded by the state.

During 2000, 2282 infants received meconium screens, but less than half (1070) were required (and funded) by the state. The expansion of the criteria had a great effect on the number of screens. During the January to June period between 100 and 127 infants per month were screened. During August to December, between 224 and 304 were screened per month. The number of screens funded by the state went from about 20 per month in the first part of the year to about 170 per month during August to December.

Of the infants meeting the selection criteria, nineteen percent (19%) were presumptively positive. Among the other infants tested, fifteen percent (15%) were presumptively positive. There have been concerns about the number of false negative results and cutoff scores for the laboratory test have been revised. Under the new methodology, the number of positive tests for cocaine is increasing. During 2000, of the 205 infants testing positive in required (funded) screening, 125

(61%) tested positive for cannabinoids (marijuana), 81 (40%) for cocaine, 18 (9%) for opiates (heroin, morphine, codeine), and none for phencyclidine (PCP). About 19 (9%) were positive for more than one drug. The breakdown by type of drug among all 389 infants who tested positive was:

Total positive screens	389	17%	Of all screens
Cannabinoids (marijuana)	227	58%	Of positive screens
Cocaine	160	41%	Of positive screens
Opiates (heroin, morphine, codeine)	32	8%	Of positive screens
Phencyclidine (PCP)	0	0%	Of positive screens
Multiple drugs	31	8%	Of positive screens

Available hospital reports indicated that 312 cases were referred for official follow-up. Among these 149 cases were referred to child protective services, 71 to First Steps, and 92 mothers were referred to treatment. These referral statistics came from individual hospital reports, while the other data in this section are from the screening laboratory. Of the 389 positive screens, only twenty-four percent (24%) of mothers were referred for treatment, and only seventeen percent (17%) of infants were referred to First Steps.

Hospital reporting and compliance with the meconium-screening program is a major concern. Only about sixty-five percent (65%) of Indiana hospitals participated in 2000. While the meconium-screening program is important and has been growing, it is not yet comprehensive and does not, apparently, provide adequate referral to services.

2.5 Prevalence of Developmental Disabilities

There is no systematic screening for developmental disabilities in children in Indiana. Applicants to the "First Steps" program administered by the Indiana Families and Social Services Agency (FSSA) can qualify if they are certified as having one delay of twenty percent (20%) in one developmental domain or two (or more) delays of ten percent (10%) each. Children can also qualify based on having particular medical diagnoses or being "at risk" due to a specified list of exposures and/or conditions. As of January 1, 2001, 5,667 children qualified on the basis of having specific developmental delays (in total, 7,879 qualified). The developmentally delayed children represented 3.38 percent of all children 2 and 3 years of age in the state (U.S. Census, 2000). This number is a lower bound estimate for prevalence since it is likely that some children who would qualify have not applied. In fact, outreach efforts in the First Steps Program have been rapidly increasing participation in recent years. As of January 1, 1999, developmentally delayed applicants totaled 3,797. That increased to 4,757 on January 1, 2000, and to 5,667 as of the first of 2001.

The lower bound prevalence rate for developmental disabilities of 3.38 percent would translate to an annual incidence rate of 1.69 because prevalence is estimated for the population of two and three year olds. To put this pattern in context, the 2001 per 10,000 is about are estimated to be half of the rate for all nine congenital anomalies traced on the map summarized in Figure 2.14.

2.6 Conclusions

It is difficult to assess the true incidence of birth defects in Indiana because of limitations in the surveillance systems. The rates of congenital anomalies (structural birth defects) are lower than in other states at least partly because of a less comprehensive screening process. Some anomalies may not be readily apparent at the time of birth so that linked claims data and records of other hospitalizations become necessary. Also, the Indiana process is "passive" in that it accepts data generated beyond its control without direct verification. A more "active" system is preferable where employees of the department are directly involved in collecting and verifying the data.

The screening process for some metabolic disorders is more active and comprehensive. It involves verifying that services and follow-up evaluations were provided to children who screen onto the Genetics Confirmed Positive Registry. This system has recently been expanded to test for more types of disorders, but covers relatively few children (about 100 per year come onto the registry).

Conversely, screenings for hearing disorders and exposure to drugs (as detected in a meconium screening) seem to be much less developed. There seem to be some problems with hospital reporting in relationship to both the hearing disorders and meconium screenings.

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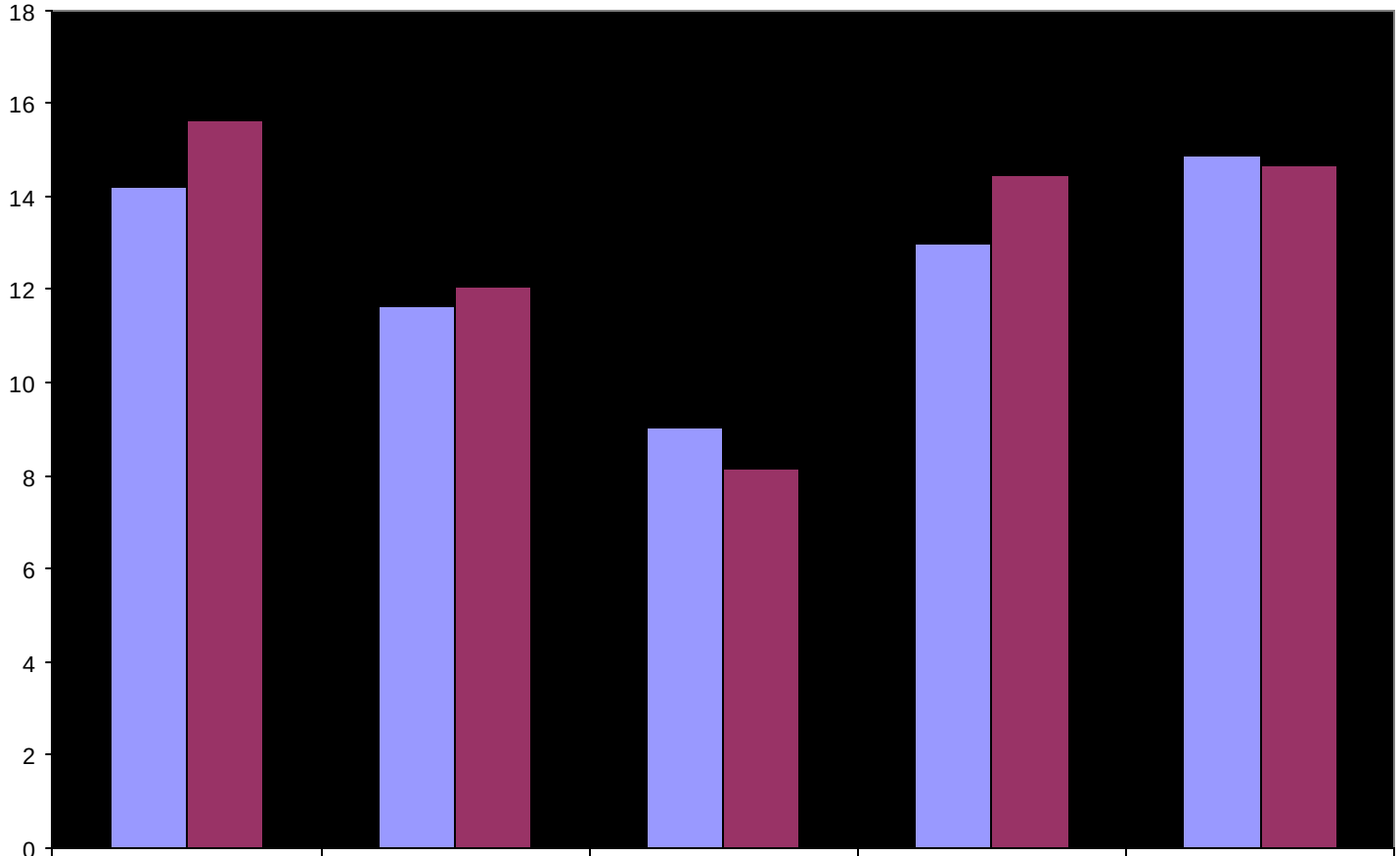
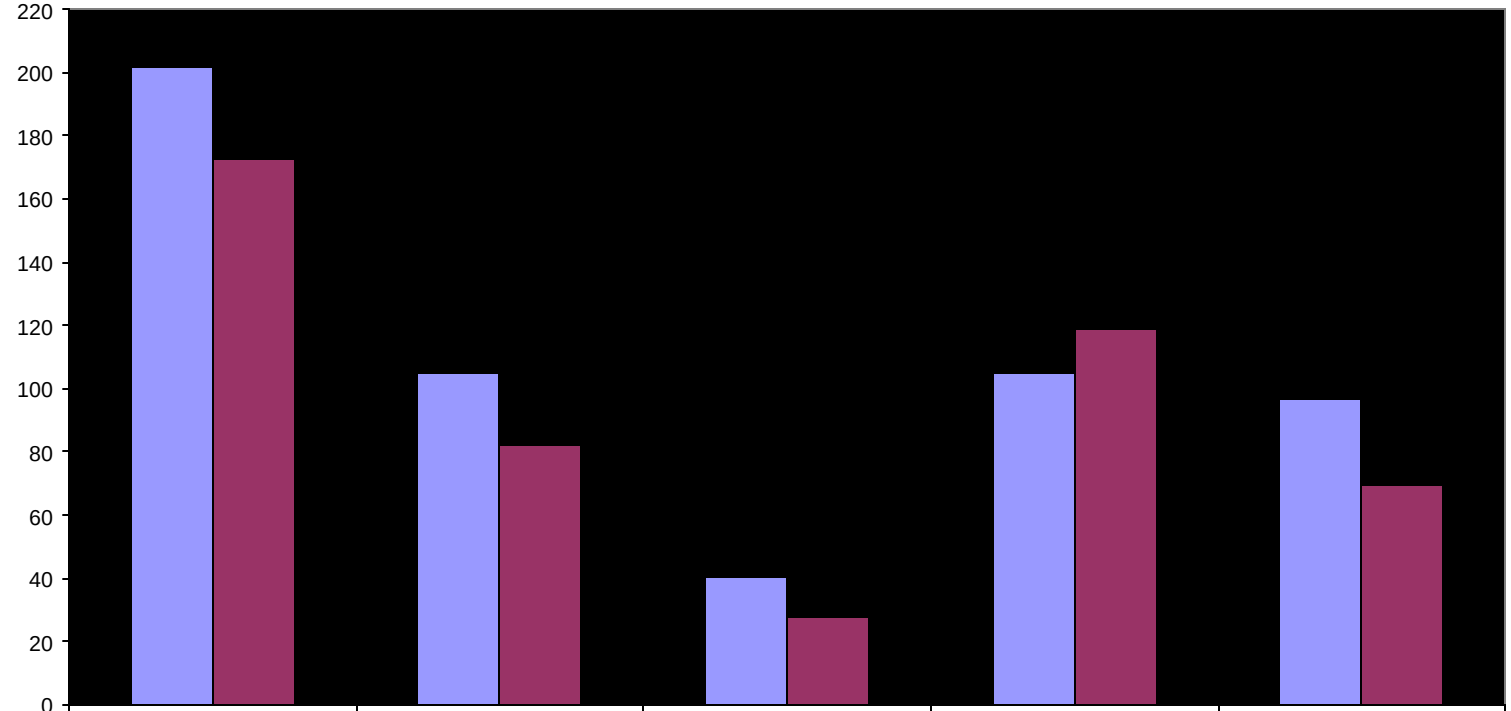
FIGURES

Figure 2.2:
Chromosomal Congenital Anomalies Per 10,000 Live Births



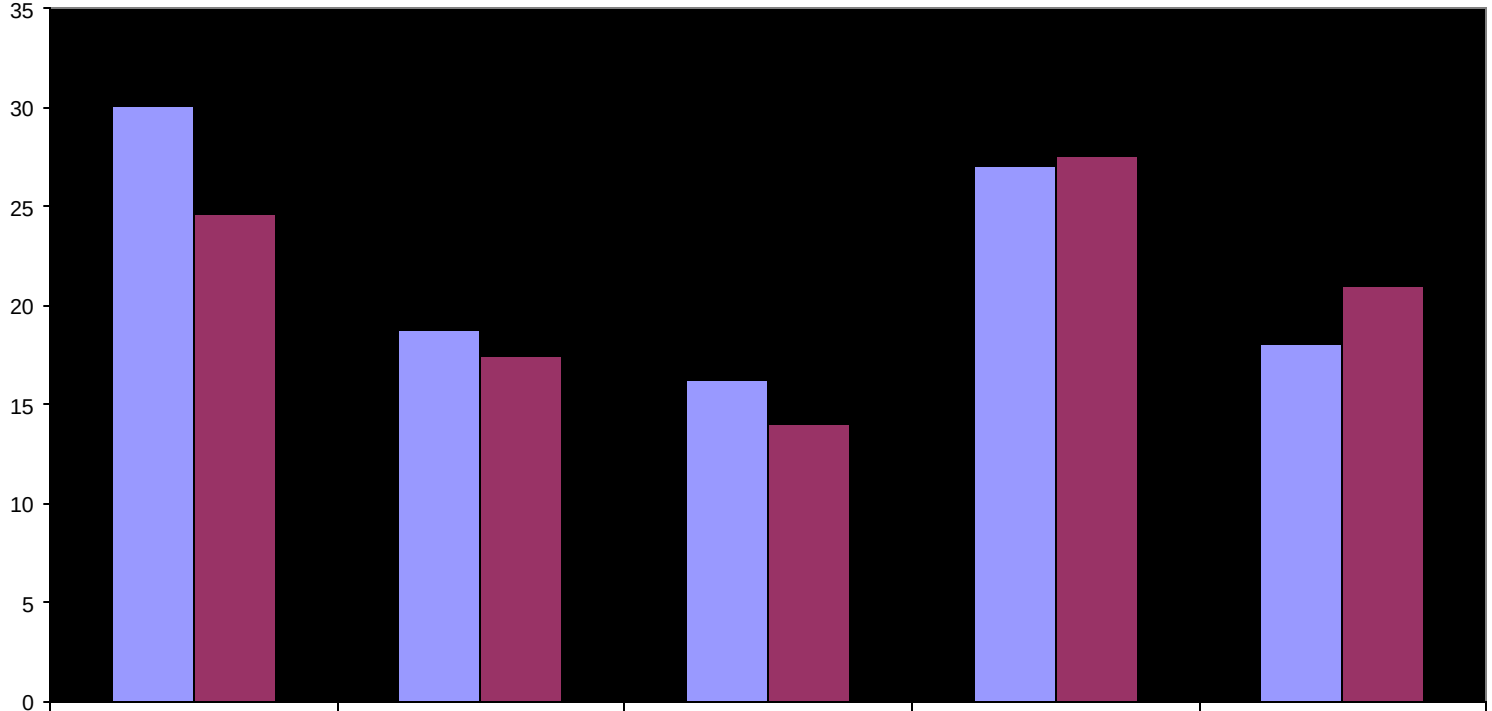
	Georgia	Illinois	Indiana	Iowa	Wisconsin
1996	14.2	11.64	9.02	12.97	14.87
1989-1995	15.65	12.05	8.15	14.46	14.65

Figure 2.3:
Circulatory/Respiratory Anomalies Per 10,000 Live Births



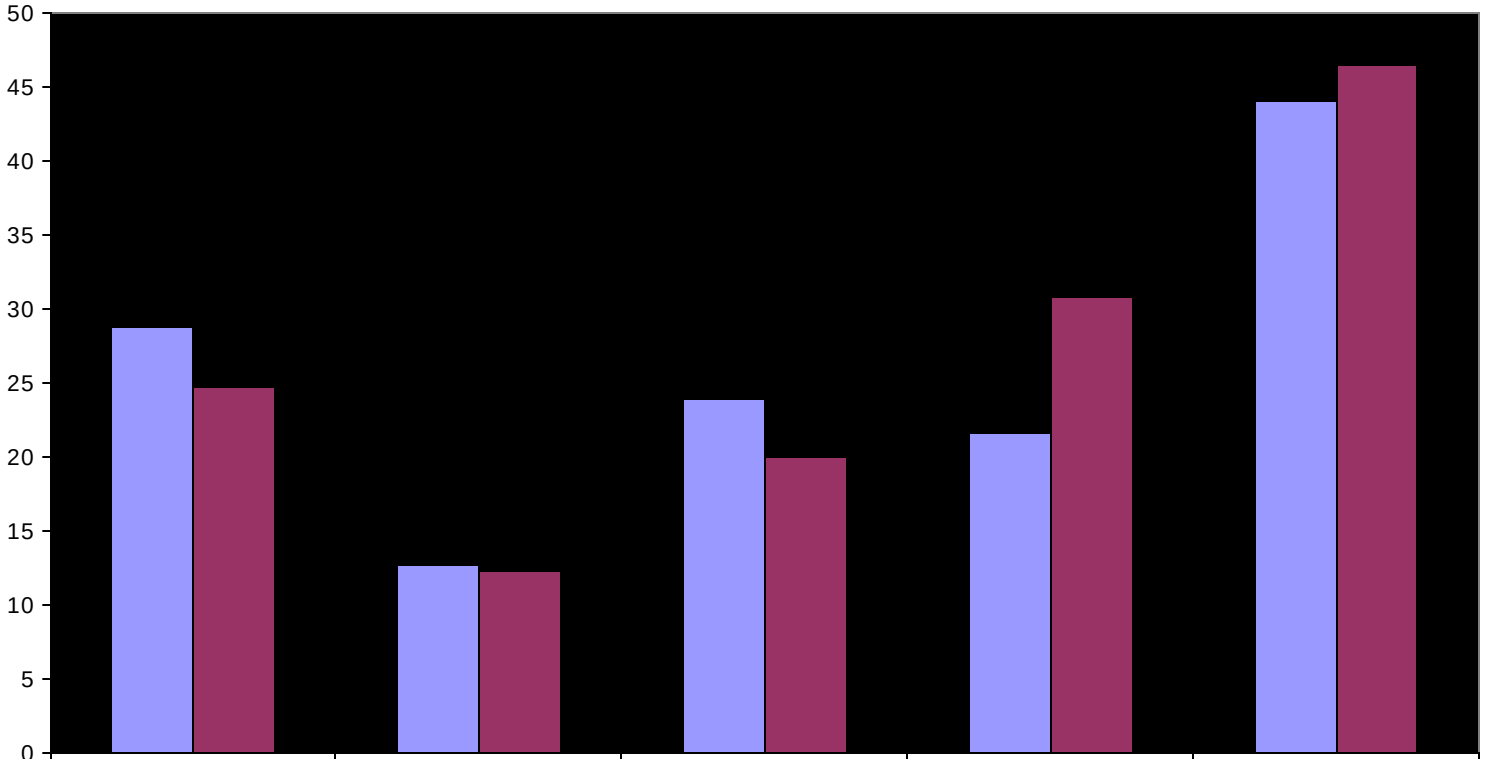
	Georgia	Illinois	Indiana	Iowa	Wisconsin
1996	201.93	104.88	40.41	104.73	96.64
1989-1995	172.67	81.87	27.79	119.01	69.21

Figure 2.4:
Central Nervous System Anomalies Per 10,000 Live Births



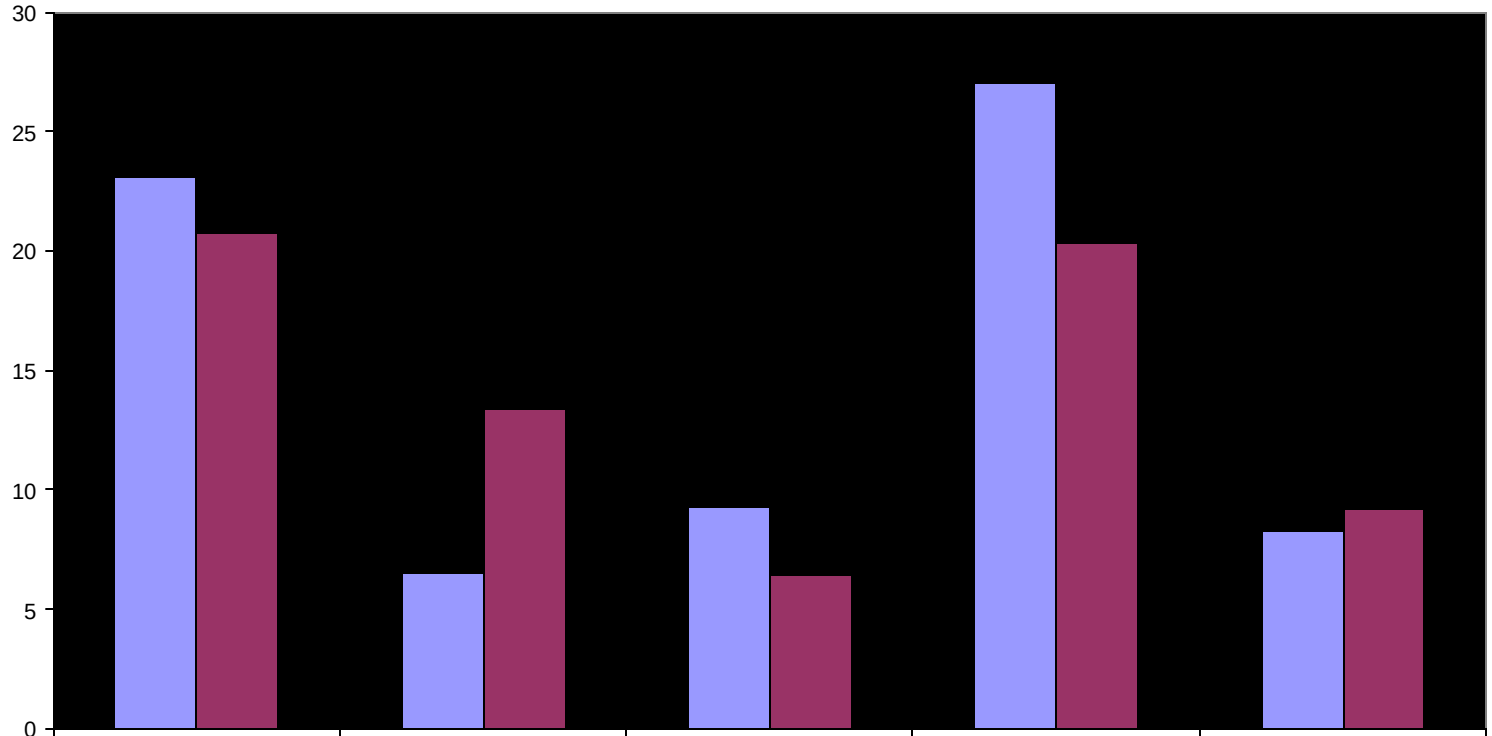
	Georgia	Illinois	Indiana	Iowa	Wisconsin
1996	30.07	18.71	16.23	27.05	18.05
1989-1995	24.62	17.4	14.05	27.52	20.96

Figure 2.5:
Cranial/Facial Anomalies Per 10,000 Live Births



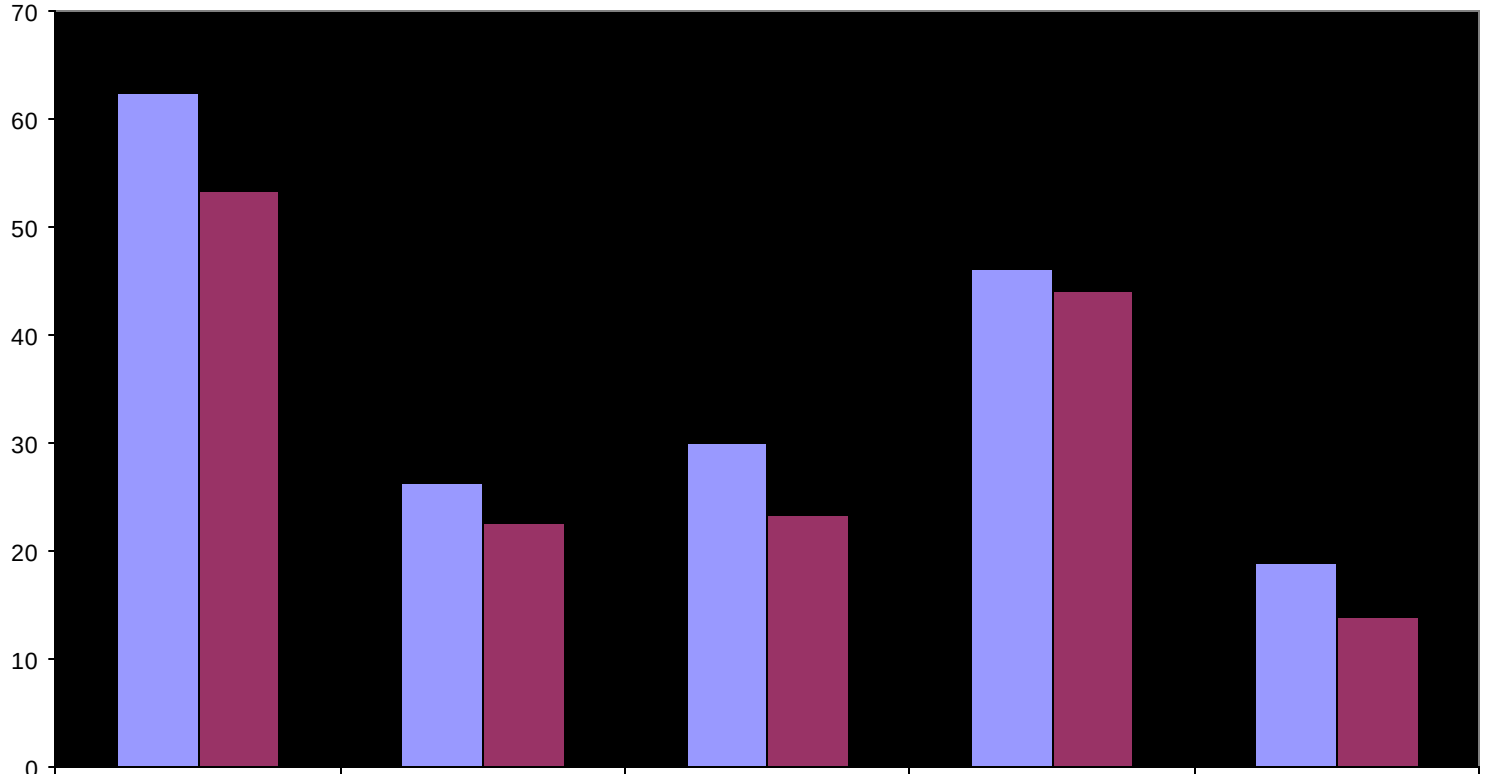
	Georgia	Illinois	Indiana	Iowa	Wisconsin
1996	28.84	12.75	23.93	21.7	44.12
1989-1995	24.74	12.23	19.96	30.78	46.47

Figure 2.6:
Gastrointestinal Tract Anomalies Per 10,000 Live Births



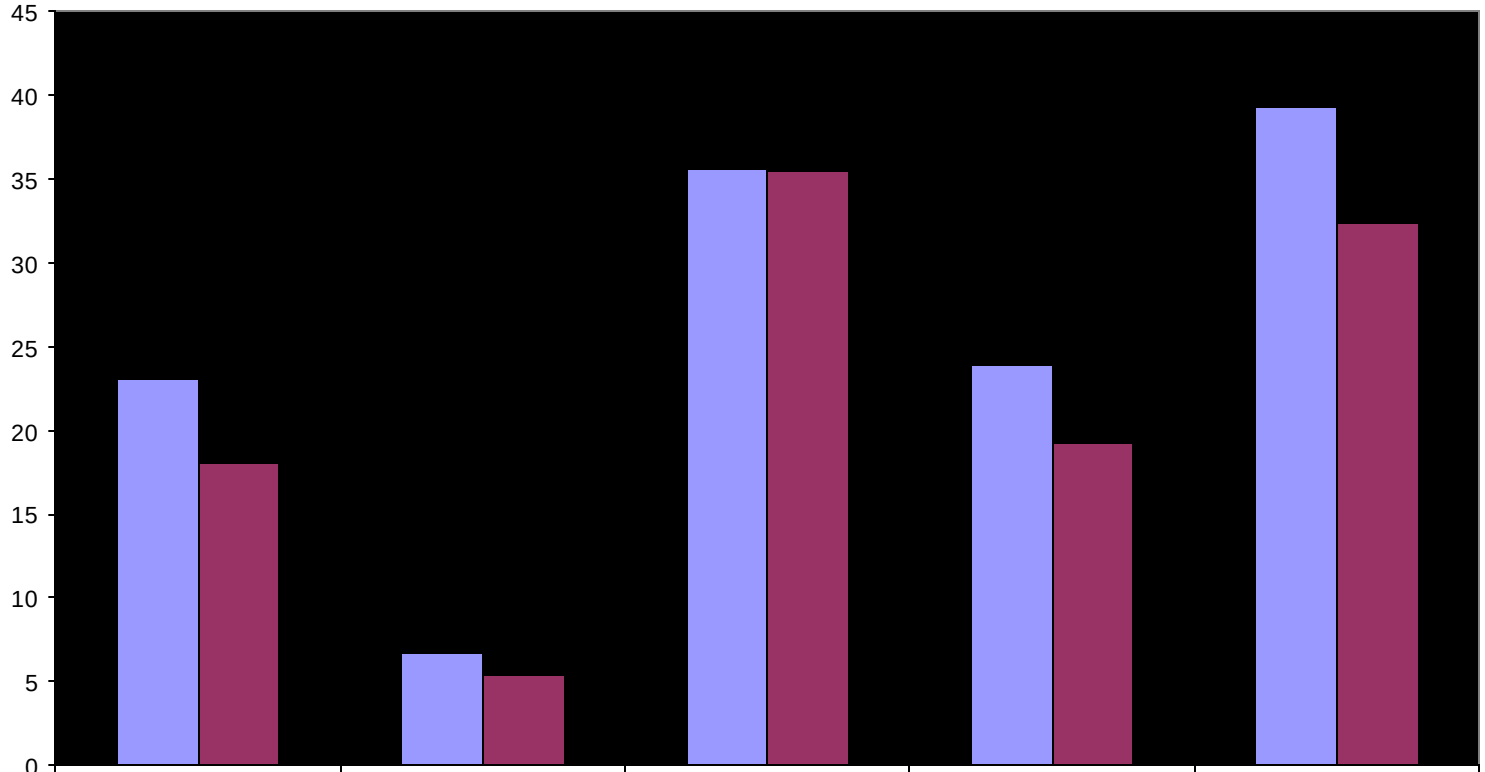
	Georgia	Illinois	Indiana	Iowa	Wisconsin
1996	23.12	6.53	9.26	27.03	8.31
1989-1995	20.78	13.34	6.45	20.37	9.17

Figure 2.7:
Genital/Urinary Anomalies Per 10,000 Live Births



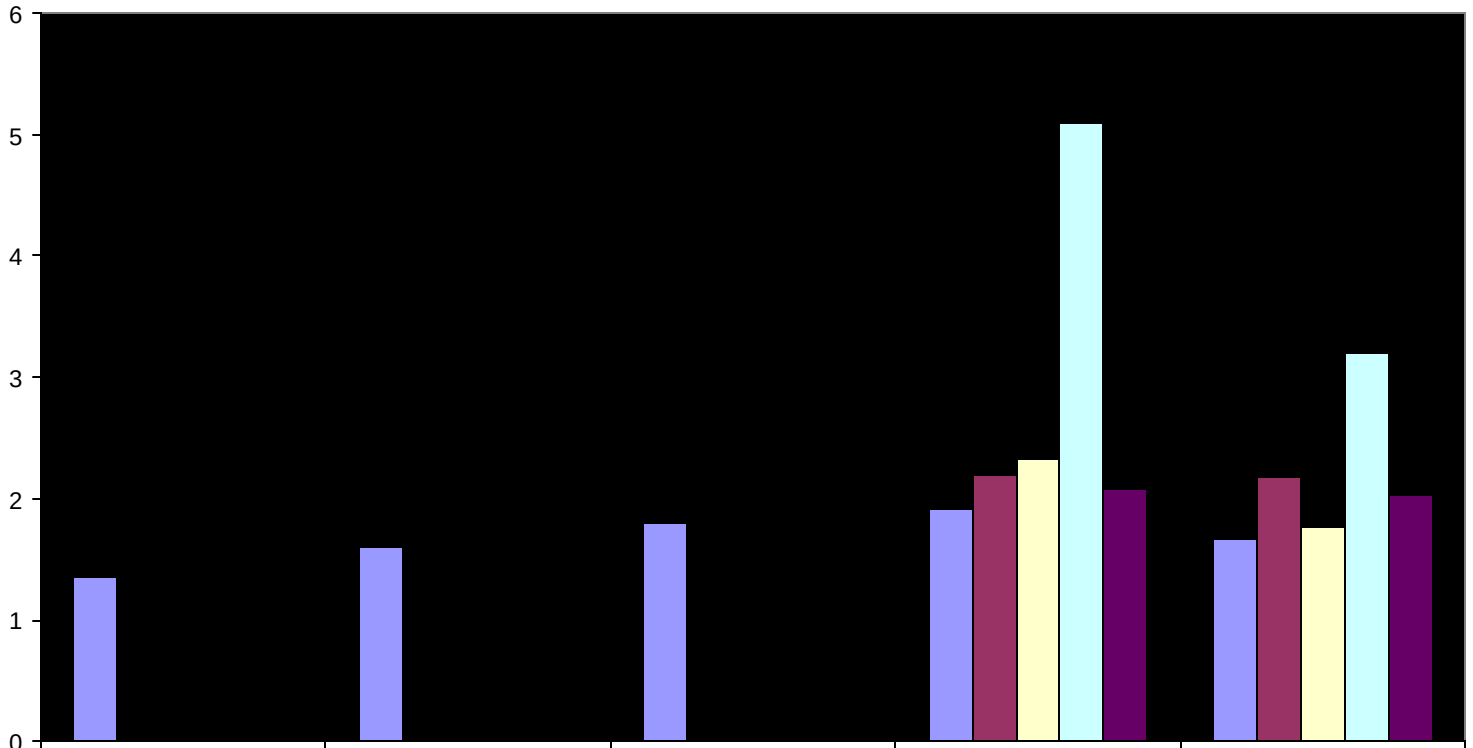
	Georgia	Illinois	Indiana	Iowa	Wisconsin
1996	62.34	26.36	30.06	46.07	18.95
1989-1995	53.28	22.48	23.37	44.06	13.82

Figure 2.8:
Musculoskeletal/Integumental Anomalies Per 10,000 Live Births



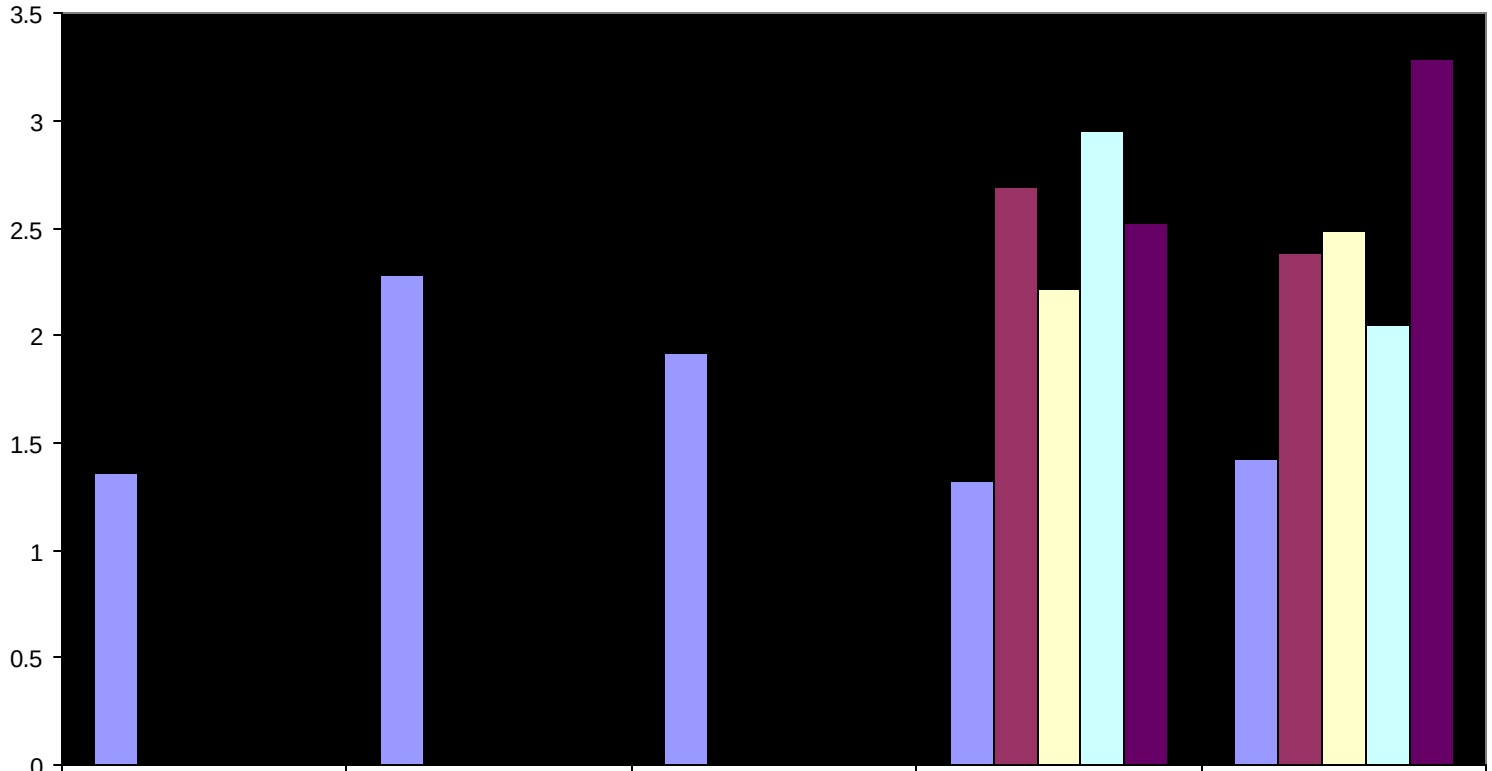
	Georgia	Illinois	Indiana	Iowa	Wisconsin
1996	23.09	6.65	35.60	23.91	39.26
1989-1995	18.09	5.42	35.47	19.3	32.41

Figure 2.9: Anencephalus Rate Per 10,000 Births



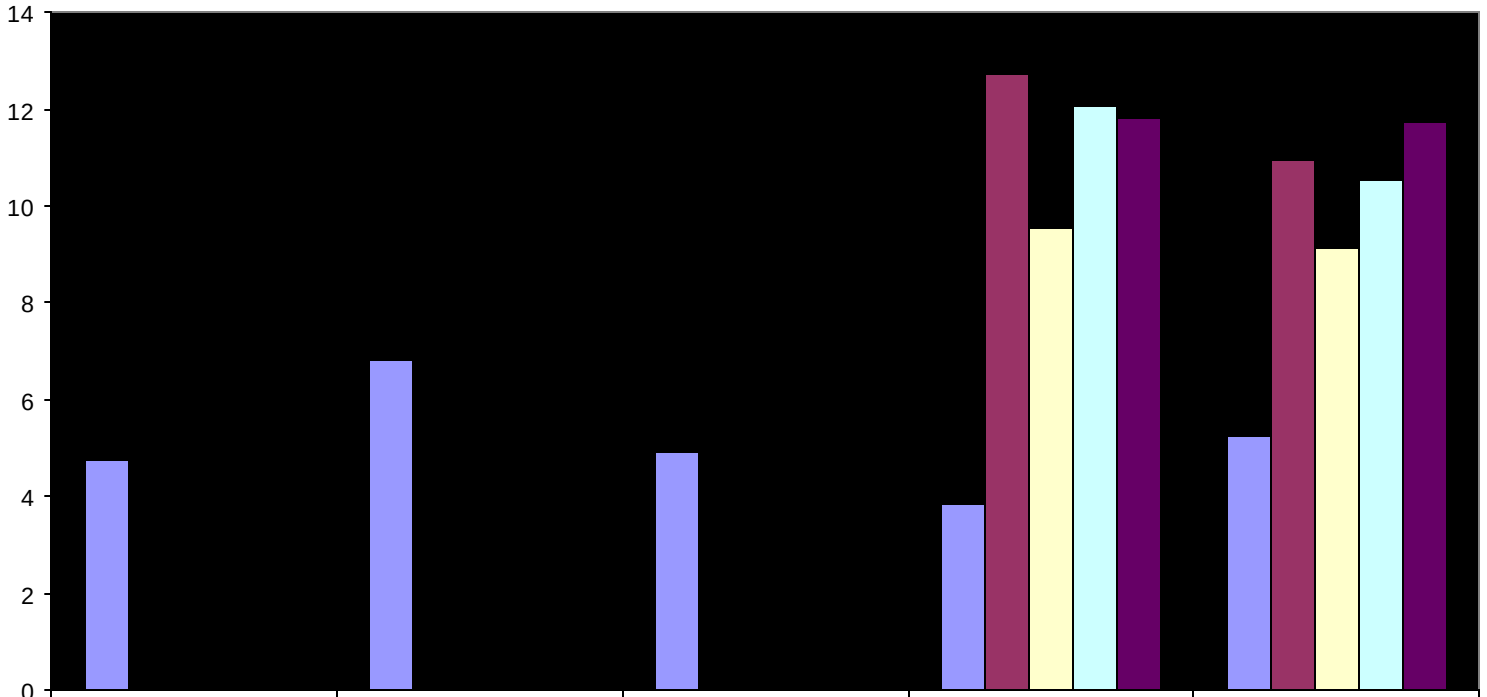
	1999	1998	1997	1996	1989-95
Indiana	1.36	1.6	1.8	1.92	1.67
Georgia				2.2	2.17
Illinois				2.33	1.76
Iowa				5.09	3.2
Wisconsin				2.07	2.02

Figure 2.10: Diaphramatic Hernia Rate Per 10,000 Births



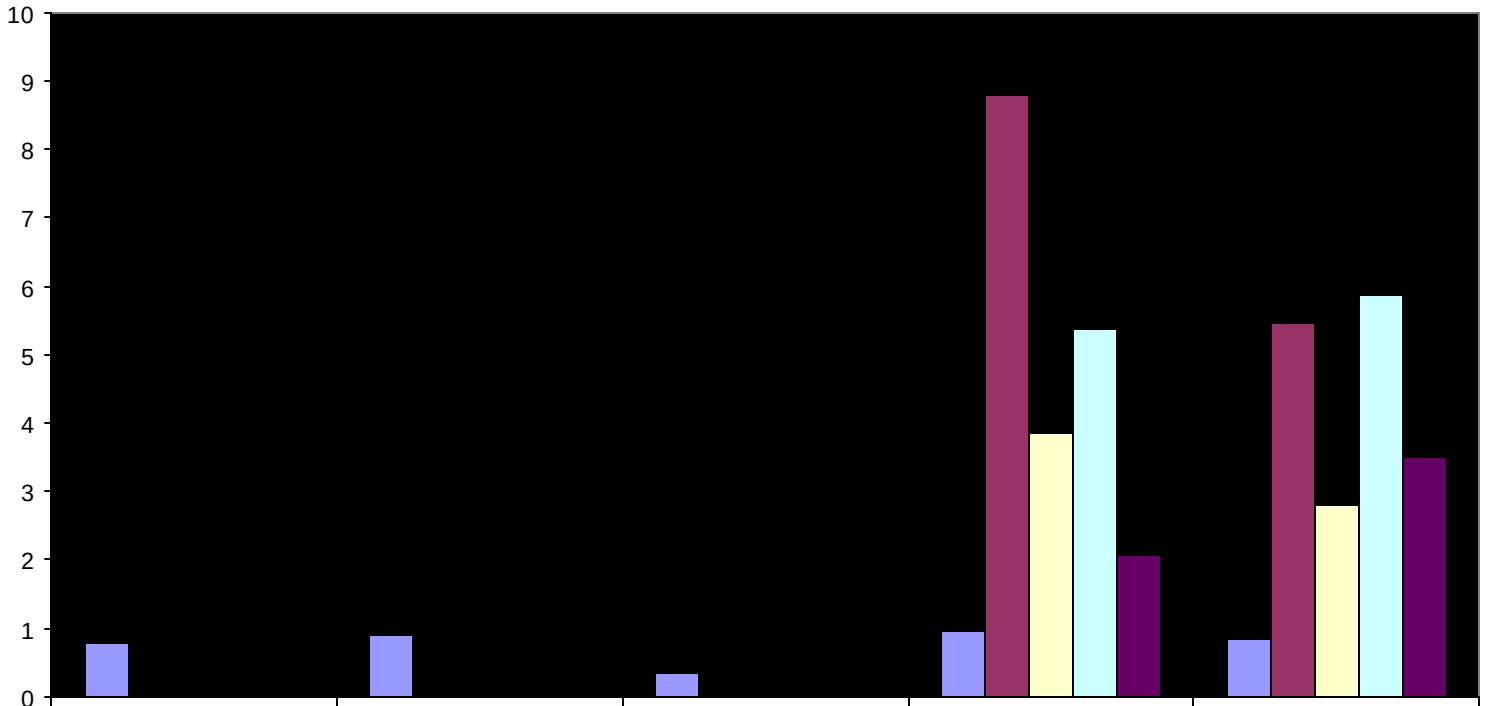
	1999	1998	1997	1996	1989-95
Indiana	1.36	2.28	1.92	1.32	1.43
Georgia				2.69	2.39
Illinois				2.22	2.49
Iowa				2.95	2.05
Wisconsin				2.52	3.29

Figure 2.11: Down Syndrome Rate Per 10,000 Births



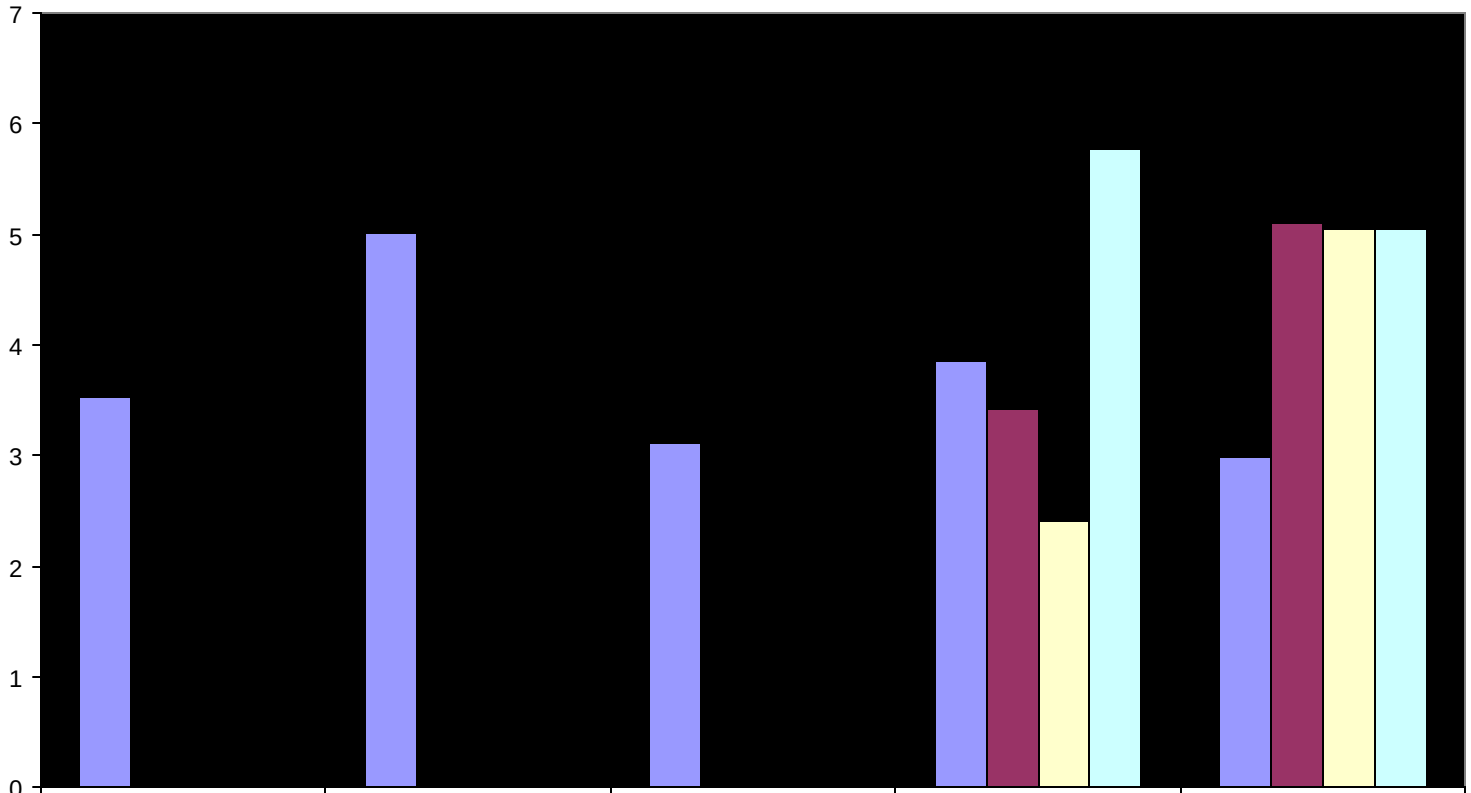
	1999	1998	1997	1996	1989-95
Indiana	4.76	6.84	4.92	3.85	5.24
Georgia				12.71	10.97
Illinois				9.55	9.13
Iowa				12.05	10.52
Wisconsin				11.84	11.72

Figure 2.12: Microcephalus Rate Per 10,000 Births



	1999	1998	1997	1996	1989-95
Indiana	0.79	0.91	0.36	0.96	0.83
Georgia				8.8	5.47
Illinois				3.85	2.79
Iowa				5.36	5.87
Wisconsin				2.07	3.49

Figure 2.13: Omphalocele / Gastroschisis Rate Per 10,000 Births



	1999	1998	1997	1996	1989-95
Indiana	3.52	5.01	3.12	3.85	2.98
Georgia				3.42	5.1
Iowa				2.41	5.05
Wisconsin				5.77	5.04

Figure 2.15: Hospitalizations in 1998 and 1999 for Congenital Anomalies (4082 Total)

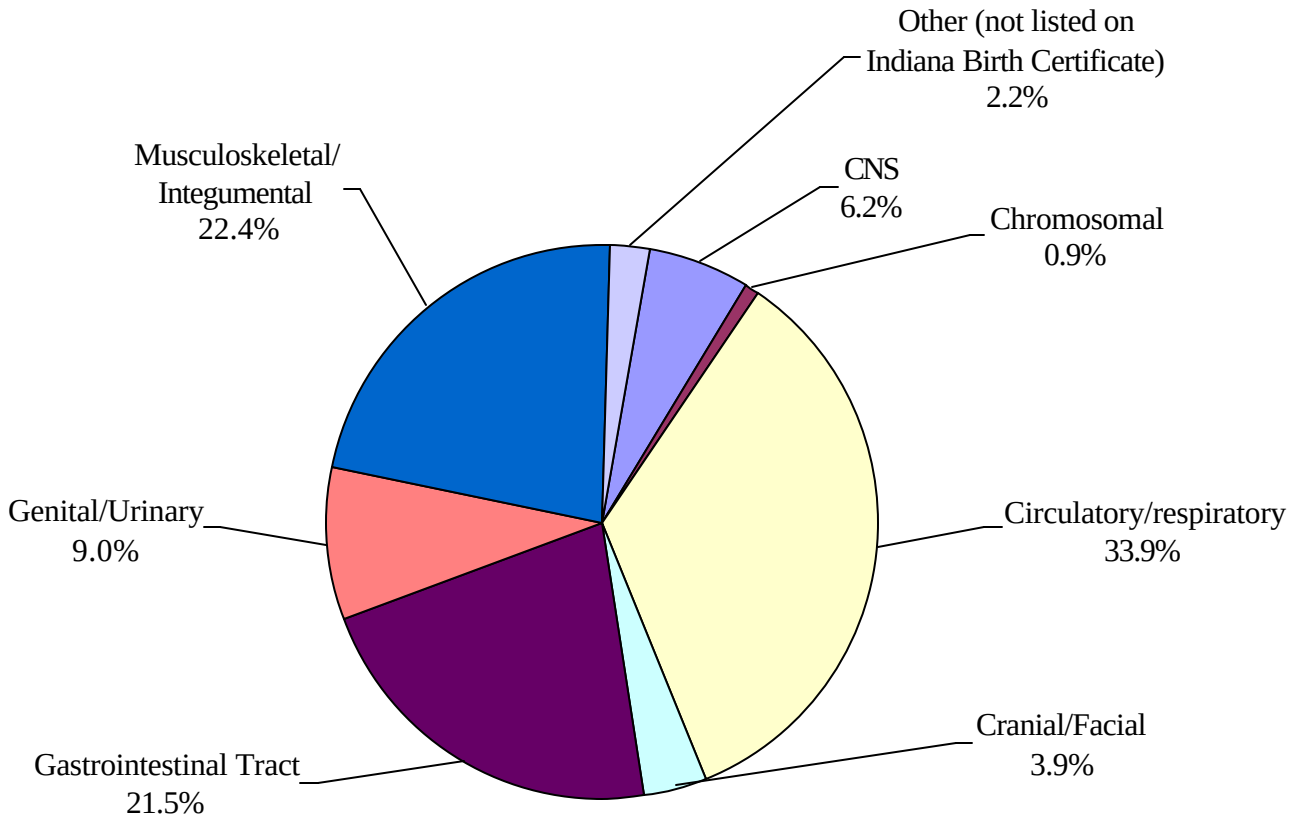
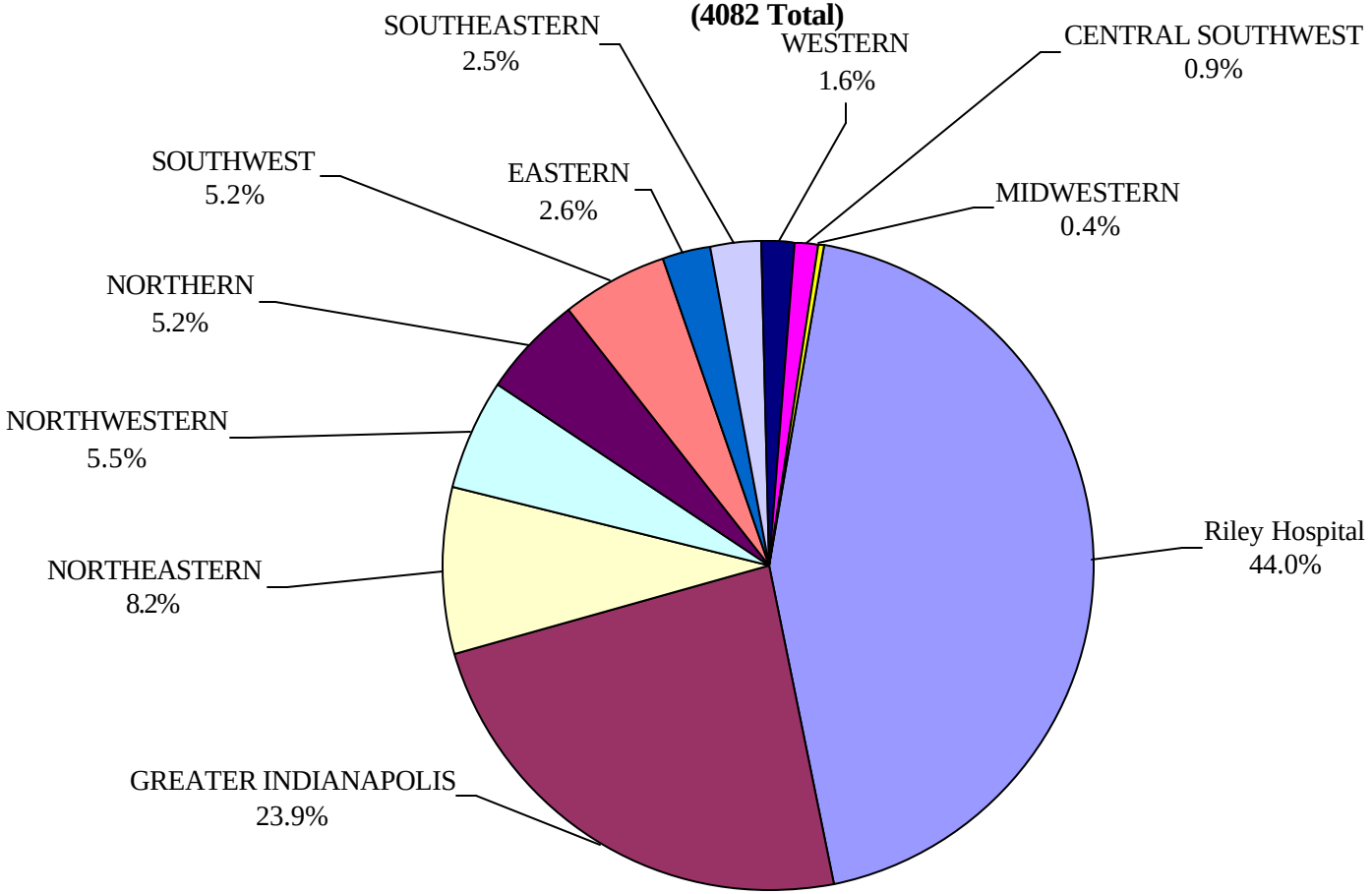


Figure 2.16: Hospitalizations in 1998 and 1999 for Congenital Anomalies (4082 Total)



3.0 SERVICE PROVISION AND GEOGRAPHIC DISTRIBUTION

The availability of genetic services, particularly in rural areas, is an emerging issue that will confront the citizens of Indiana during the coming decade. The force driving Indiana's need to fully explore the availability of genetic services is due in part to an increased recognition of the prevalence of genetic disorders through state mandated pre-natal and early childhood screening programs. Likewise, the rapidly increasing number of government and privately funded research programs has exposed the limits of service providers' expertise in treating the complex issues associated with genetic disorders (Khoury & Burke, 2000). It therefore follows that a comprehensive understanding of the availability and strength of Indiana's genetic-related service providers is not only needed but also essential (for the state) in formulating a long-term strategy to equitably distribute financial resources and effectively combat the rising number of reported genetic disorders.

Indiana has a largely metropolitan populace (64% urban, 36% rural) that is still experiencing greater growth in the urban areas with rural inhabitants relocating to the more centralized "big city" setting. The health care system in Indiana appears to mirror the demographics of the population, with the more refined and specialty health care services offered in the larger metropolitan areas with the rural areas limited to the basic health care providers.

Although other agencies have produced insightful studies and conclusions regarding particular components of Indiana's genetic health care system, little research has been undertaken to comprehensively analyze the present genetic health care services in terms of its relevance to known prevalence rates, assessment of statewide genetic medical professionals, genetic service providers, network of provider institutions, and underserved/over served areas.

It is therefore the scope of this section to examine, analyze, and determine the availability and distribution of Indiana's genetic service providers in terms of a modified Council of Regional Networks for Genetic Services (CORN) assessment protocol. In addition, several geographic mapping techniques are employed to highlight the distribution of services providers across the state.

3.1 Methodology

Various methodologies are commonly employed to ascertain the strength of health service providers in a particular county or given population. Some of the more common include the Health Manpower Shortage Area (HPSA) or the Medically Underserved Populations (MUP) method. Though either of these would be sufficient to address the genetic services in the state of Indiana, the Council of Regional Networks for Genetic Services (CORN) has specified particular criteria needed to assess a state's genetic services and ultimately evaluate a particular state plan. This particular component of the needs assessment will address the availability of the community services with the CORN evaluation criteria as a guideline for analysis.

Specifically, the community assessment examines Indiana's genetic services in terms of the distribution of health care professionals, developmental services providers, licensed physicians, and genetic services offered by private/publicly funded genetic clinics throughout the state. Data

collection for the community assessment piece was conducted over a two-month period from personal interviews and various data sets provided by the Indiana State Department of Health and the Family and Social Service Administration. In some cases, the sources produced inconsistent numbers. When inconsistencies occurred, the personal interview data were utilized as the most current interpretation of service availability.

To assess the infrastructure of Indiana's genetic-related clinics, a simple data collection instrument (Figure 3.1) was designed using CORN's "Guidelines for Genetic Needs Assessment (GCGS)" (See Appendix C) as a baseline and then further segmented into four sub-categories which include genetic services offered at the clinic site, the number of genetic-related staff employed or contracted out by a particular genetic service network, the type of educational activities offered across the state, and the types of population screenings offered at a particular clinic. Each of the instruments sub-categories was based on GCGS criteria and definitions (See Appendix C: GCGS, page 18).

Contact was made with each of the core ("planet") genetic service sites by phone and interviews were conducted with a "knowledgeable" representative(s) of that particular institution. The term planet is used through this section of the report to designate a core location. Contact was initiated only with those institutions that met the CORN GCGS criteria, and subsequently were listed in the "Genetic Service Providers-Yellow Pages." Any information about a "satellite" location was acquired through the planet location. Tables 3.1, 3.2, & 3.3 show the compilation of the services offered, number and type of genetic staff by location, educational activities offered by region, and type of screenings offered at each of the clinics.

To better understand the dynamics of the genetic clinics and service coverage capability within the state, two maps were plotted, designating each of the networks with a unique color. The planet clinic was displayed with a large square and their satellite clinic(s) with a circle. Figures 3.2 & 3.3 show the networks superimposed on a map of the rates of congenital anomalies (from Figure 2.14). This analysis allows a general impression of how the location of service sites relates to areas of need. These genetic services networks and their staffing are discussed in detail in Sections 3.5 and 3.6.

To understand the distribution of other health service providers across the state, data sets provided by the Indiana State Family and Social Service administration (FSSA) were prepared for geographical analysis in the Arcview GIS (V 3.2) software package. The data set comprised over 3,400 cases of service providers that are routinely tracked by FSSA and categorized as those who provide services for developmentally delayed, speech delayed, or counseling services. The count of providers by category is shown in Table 3.4. Because of the large range of professions (N=38), they were condensed into 12 (N=12) similar sub-classifications for clarity and ease of analysis.

These 12 sub-classifications (shown in Table 3.5) are as follows: Speech and Hearing [Speech and Hearing Therapists], Alternative Therapy [Massage Therapists], Developmental Services [Developmental /Educational Associate, Developmental Assistant, Developmental/ Educational Specialist], Transportation [Ambulance, Bus, Common and Non-Common Transports], Medical Physicians [other than those located in Table 3.6, i.e. ENT, Surgeon, etc.], Language Interpreters,

Nurses [Licensed Practical Nurses (LPN), Registered Nurses (RN)], Allied Service Providers [Physical Therapists (PT), Occupational Therapists (OT), OT Assistants, PT Assistants, Rehabilitation Services], Counseling Services [Marriage and Family, Psychiatrist, Psychologist, Social Worker-MSW], Registered Dietician with a specialty in genetics, Service Coordination [Service Coordinator, Service Coordination Associate], and vision specialists. The distribution of these services is discussed in Section 3.4.

Also important to understanding Indiana's genetic needs is the distribution of physicians (who may encounter a genetic-related case) throughout various regions of the state. Based on the GCGS, data for the number of physicians with the following specialties were indexed using a frequency table by the number in each county. Each county was further classified, based on the Indiana State Census information, as either rural or metropolitan (See Table 3.6). The distribution of physicians is discussed in Section 3.3.

3.2 Population and Demographics

The need for genetic services directly coincides with population growth and demographic projections for Indiana and will ultimately affect how accessible services will be for genetic disorders. Geographically Indiana ranks 38th in size (area) and is one of the top twenty in population in the U.S., with a total of 5, 900,000. Projections for Indiana indicate growth through the year 2020. Indiana currently ranks as the 28th fastest growing state in the nation since 1990. However, Indiana's share of the nation's population continues to decline, from 2.56 percent (2.56%) in 1970, to 2.42 percent (2.42%) in 1980, to 2.23 percent (2.23%) in 1990 and to 2.18 percent (2.18%) in 1999.

In each year since 1990, Indiana has seen net in-migration. With more persons moving into the state than moving out, this pattern represents a significant reversal of the out-migration experienced in the 1980s. This recent in-migration, combined with natural increase, has resulted in relatively rapid population growth for the state of Indiana in the 1990s. Of the net population increase of 399,000 persons since the 1990 census, about seventy-three percent (73%) were the result of more births than deaths and about twenty-seven percent (27%) due to net in-migration.

Of the 92 counties in Indiana, the ten largest in context of population are: Allen, Elkhart, Hamilton, Lake, Madison, Marion, Porter, St. Joseph, Tippecanoe, and Vanderbugh. Elkhart, Hamilton, Johnson, Madison, and Vanderbugh counties are all expected to see substantial increases in population by the year 2020. Counties projected to decline in population include Delaware, Grant, and Vigo.

3.3 Primary Care Physicians

Primary Care Physicians are essential as a primary point of entry for families and patients with birth defects and genetic conditions. The primary care physician offer consultation and referral to appropriate services to their clientele. The distribution of primary care physicians by county (Table 3.6) indicates that almost ninety percent (90%) of the physicians (Family, Internal, General Practitioner, OB/GYN, and Pediatrics) practice and live in metropolitan areas, with almost half of the physicians located in the five largest cities of Evansville, Ft. Wayne, Gary,

Indianapolis, and South Bend. Slightly more than ten percent (10%) practice in the rural areas that comprise a total of fifty-five counties. Arguably, a majority of the population lives in these five large population centers. Individuals requiring services that live in the rural areas are at a clear disadvantage in that they may have to travel up to two hours to reach a large population center.

Individuals residing in the southeastern portion of the state are particularly disadvantaged, as they are required to travel up to an hour (60 miles) to reach a population center large enough to support a variety of primary care services. In many of the counties, such as Ohio and Switzerland, there are no reported Pediatric, OB/GYN, or Internal physicians practicing or the county. The residents in these areas have little choice but to seek primary care services in Ohio, Kentucky, or travel to Jeffersonville.

Other counties that are underserved with primary care physicians include Ohio, Spencer, and Union, with only one physician reported in the county. In some cases the single physician servicing rural counties in Indiana is a non-board certified general practitioner. Sixty percent (60%) of the rural counties have fewer than 10 licensed physicians practicing in their county at any given time. Of the remaining 20 larger counties designated as rural, only three have more than 20 physicians. The maximum among any rural county (as in Marshall county) has 30 physicians. The area remains inadequately served with disproportionate numbers of physicians practicing in family medicine (20), three (3) pediatricians, and only one OB/GYN.

Of concern are the rural counties and particularly smaller ones that require more than one (1) hour travel time to a county that is larger and/or has access to necessary resources. These counties have inadequate primary care resources where genetic screening might occur. Other regions of concerns include the area south of St. Joseph County and north of the Indianapolis Metropolitan Statistical Area (MSA) and the region south of Indianapolis and northeast of the Evansville MSA where there is a gap in number of service providers that can provide primary care without residents having to travel up to 60 minutes for services.

3.4 Developmental Services

The infrastructure for treatment and guidance in daily living skills for individuals who suffer from genetic conditions are derived from developmental service providers. These include specialties such as physical therapists, occupational therapists, and psychiatric services. To ascertain the availability of such services in the various regions of the state, GIS plots were used to better understand the availability of providers throughout the state. Some of the findings confirm the understaffing of services in the rural regions of the state.

The use of Audiologist and Speech Specialists is by far the most widely utilized service by developmentally challenged children (See Figure 3.4). With over 900 unique providers in the state, there is a clear distribution pattern with the greatest number of specialist located in and around the Indianapolis metropolitan area, with the Hamilton county area having the second largest number of service providers. Areas that rank just below the Indianapolis area in relationship to the number of providers include Ft. Wayne and Delaware County, where Ball State University is located.

Many of the larger cities average between six to ten providers per zip code, with rural counties having only one to two providers per county. Many of the smaller rural counties do not have any reported service providers with in the speech and hearing specialty thus requiring travel to a different county for services.

In particular, residents in Warren, Fountain, Benton, and Montgomery counties travel up to sixty miles to obtain services in either Tippecanoe or Indianapolis. Equally problematic are those who reside southwest of Indianapolis in Owen, Sullivan, Clay, Putnam, and Davies county where a total of seven speech specialists service all five counties. The triad of counties to the southeast, Dearborn, Ohio, and Switzerland, have no providers and the residents must travel the distance of two counties to receive services. Within the northeastern counties a medium-sized city is situated within sixty miles; however, speech and hearing services are unavailable.

Similar to the patterns identified for speech and hearing services, the Counselors, Psychiatric, and Psychological Services (Figure 3.5) are located in the central metropolitan areas, with residents in the rural counties traveling up to sixty miles for services. The term "Counselors" is defined by the CORN guidelines as a "health professional with a Master's degree who is academically and clinically prepared to provide genetic counseling services to individuals and families seeking counseling information about the occurrence, or risk of recurrence, of a genetic condition or birth defect" (Appendix C, page 23). The same pattern is present for a limited number of services in the triad of counties to the southeast (Dearborn, Ohio, and Switzerland) where no counselors are located. Residents must travel across as many as two counties for services. Other areas of concern include the central-western portion of the state in Warren and surrounding counties where travel to Tippecanoe is required for services. Counseling service providers are located in largely metropolitan areas, often leaving many of the smaller counties underserved. There are definitive underserved areas to the north of Ft. Wayne and in the southeast region, where the only care center is in Jeffersonville or at the University of Louisville.

The patterns of Developmental Service Providers and Service Coordinators (Figure 3.6) are distributed similarly as other resources with larger metropolitan areas containing the majority of service providers. The Developmental Service Providers and Service Coordinators are contracted for by the state and responsible for developmental assessments, service coordination, and case management. In the case of service coordinators, the professionals travel to each family to develop service plans. Thus, although the coordinators are not physically located in each county, the service is delivered individually to each family.

Physical Therapists (Figure 3.7) that serve children with developmental problems are clearly concentrated around the Gary and Indianapolis region with a disproportionate number located in the IU/Clarian ZIP code. Other areas that have substantial numbers include the southeast area around Jeffersonville. Medium-sized cities in the state, such as Lafayette and Crawfordsville, have at least one PT who works with developmentally challenged children. But on the whole, it is clear that those who live in the rural areas must travel at least through one county and perhaps two to seek services.

Specialties in Allied Medicine Providers and Nurses (Figure 3.8) who work with developmentally delayed children comprise a limited number of providers in the state and are concentrated primarily in the major or secondary metropolitan areas. One finding that is not surprising is the use of transportation by individuals seeking developmental services. Those counties that have greater proportional use of transportation are rural counties with limited number of primary care physicians (distribution not shown). Interpretation services, primarily for Spanish interpreters, have greater utilization and availability in larger metropolitan areas and little if any use in the rural areas of the state which is possibly a reflection of availability (distribution not shown).

Areas such as Eye/Vision, Dietary, Medical Specialists, and Alternative Medical Specialties, have inadequate data available to draw qualified conclusions with respect to any particular region or area.

3.5 Genetic Clinics and Networks

The nucleus for genetic services (in the state) is the individual genetic service network and its providers. Using the data from the telephone calls to the various genetic service providers and plotting the information with GIS, it is easy to ascertain areas that are adequately covered and those that may be underserved.

Without question, the largest network that serves the most people is the network of Indiana University and Clarian (primarily Riley Hospital for Children) (IU/Clarian). The network (shown in Figure 3.3) is financially supported through state grants, federal funding, and private donations. It serves large population centers of South Bend in St. Joseph county (north) and Evansville and Vanderburgh county (south), while serving the smaller population centers in Vigo, Monroe, and Jefferson counties. Services are also provided at the Indiana University Prenatal Diagnostic Center located in Terre Haute. They offer the greatest number of support services in the state with a team of service providers that travel to each of the four satellite sites. On staff are five (5) clinical geneticists, two (2) cytogenetists, two (2) bio-geneticists, 13 cytogenetic technologists, eight (8) genetic counselors, and one (1) diet specialist. As one of the ten largest hospitals in the U.S. and the largest hospital in Indiana, they are able to combine resources from the Indiana University School of Medicine and other areas of the hospital to provide the most complete network in the state.

Other networks located in the Marion County area (see Figures 3.2 and 3.3) include the Center for Prenatal Diagnosis (CFPD), St Vincent's Hospital, Community Hospital, and the smallest in the state, the Indiana Center for Prenatal Diagnosis (ICFPD), which provides fertility services to clientele who have private insurance coverage or are self paying. Only CFPD has satellite offices (located in Lafayette and Evansville). All three appear to be medium to small genetic clinics serving the population base in central Indiana (with the exception of Evansville). Staffing is limited to no more than one (1) Ph.D. geneticist, a few cytogenetic technologists and OB/GYN's.

The Aegis network is a privately funded/not-for-profit clinic based in Monroe county (shown in Figure 3.2). They have two satellite sites and are located in Linton and Bedford. The staff is

limited to one (1) genetics counselor, one (1) OB/GYN, and one (1) diet specialist. Services include all six of the CORN standard services, and they offer folic acid and teratogenetic education activities. As well, both prenatal and family screenings are offered at all three sites, but they do not have any of the cytogenetic, biological, or molecular laboratory services on-site.

Geographically, the Clarksville network (shown in Figure 3.2) has two satellite sites located in Jeffersonville and Corydon. A third will be in Paoli and is projected to be operational in November, 2001. The staff supporting this network originates from the University of Louisville and is able to provide a substantial number of services through their planet and satellite sites. Included in their services are three (3) clinical geneticist, and two (2) genetic counselors. They offer all six areas of the CORN recommended services and provide folic acid and teratogenetic education services. All screening and labs are outsourced to the University of Louisville.

The Gary Network has three satellite sites located in Lake County, Laporte, and South Bend (shown in Figure 3.2). On staff are one (1) clinical geneticists, one (1) Ph.D. Cyto-Geneticist, and one (1) cytogenetic technician. Obstetrician, Gynecologist, and/or Perinatologist needs are outsourced to various hospitals in the city and in the greater Chicago region or by the respective service in the location; however, it is not supported by the network. Gary offers folic acid and teratogenetic education, has an on-site cytogenetics laboratory services, and offers prenatal and newborn screenings.

The network serving the northeast portion of the state is anchored in Ft. Wayne and has two satellite offices, one in St. Joseph and one in Delaware County. Their strength is clearly the seven (7) nurses that have a specialty in genetics coupled with one (1) genetics counselor, one (1) clinical geneticist/cytogeneticist, and one (1) OB/GYN. In the Fort Wayne network all the educational services identified by CORN are offered, and they have on-site cytogenetics laboratory services.

A small network located in the south-western portion of the state is St. Mary's Regional Genetic Services Center located in Evansville. The staff is limited to one (1) genetics counselor provided by Indiana University Medical Genetics, one (1) OB/GYN Perinatologist, one (1) genetics counselor, and two (2) diet specialist available on a consultative basis from St. Mary's Hospital. The OB/GYN Perinatologist and genetics counselor provide services at the Center for Prenatal Diagnosis Evansville clinic.

3.6 Genetics Staff

When staff is plotted by service network, it is clear that most of the areas in the state have access to clinical geneticists, genetic counselors, and OB/GYN's (See Figures 3.9, 3.10, & 3.11). All of the networks have at least one clinical geneticist in the planet and satellite site(s). The largest provider is the IU/Clarian network that serves most of the major metropolitan areas of the state. However, there are regions that appear to be underserved and require at least one to two hours travel time to reach care (See Figure 3.9). These include two areas 1) the north-central portion of the state and 2) the southeastern region extending to the Ohio border. Oddly enough, Monroe County is an over-served area, with access to six (6) geneticists in an area with less than 60, 000 population.

The distribution for genetic counselors (Figure 3.10) mirrors that of the clinical geneticists, with greater number of counselors available in the five larger metropolitan areas. Subsequently, residents in the smaller areas and rural locations travel thirty (30) minutes to one (1) hour for services. However, the north central and the southeastern part of the state are understaffed. The genetic specialist OB/GYN staff is distributed in many of the same locations. Exceptions to the distribution pattern are Gary and the CFDP network, where no OB/GYN staff is reported (See Figure 3.11). However, Gary and the CFDP networks outsource the OB/GYN services.

Specialties, such as dieticians and cytogeneticists, are sporadically distributed throughout the state with only one network covering a specialty leaving the remaining portion of the state underserved. In relationship to cytogeneticists, only IU/Clarian and Community Hospitals report having such professionals on staff.

3.7 Educational Programs

Educational activities (Table 3.3) aimed toward medical practice clientele are readily available in most networks including the primary and the satellite or outreach sites. The educational programs include folic acid and teratogenetic issues. Some networks and their satellite sites provide valuable clinical experience for those undergoing medical education training and therefore facilitate the education of future practitioners.

A crucial resource available to existing practitioners is an education program entitled "Genetics and Your Practice." The program is directed toward the medical community in the state of Indiana and was established in 1999 by the Indiana State Department of Health. The curriculum is offered to physicians, health care agencies, and auxiliary medical personnel. When the program originated, a geneticist and genetic counselors traveled throughout the state providing material and education in relationship to genetic issues and care. Currently the education program is available upon request.

3.8 Conclusions

Several patterns can be observed when mapping the service delivery structure of the state. Mapping the prevalence rates of various chromosomal anomalies verified that the heaviest concentrations of some problems with high rates were not in the urban areas (See Section 2, Figure 2.14). Figures 3.2 and 3.3 superimpose existing networks on the prevalence data illustrating some high prevalence rates in areas distant from core services. A high rate in a sparsely settled area does not necessarily translate into adequate raw numbers of clients to support a wide array of services. Nonetheless, the coverage available to some areas of the state does appear to present access challenges and results in lengthy traveling distances to obtain necessary services. The observed patterns can best be summarized as follows:

* It can be argued that primary services are limited in the rural counties.
* Resources are shared between the smaller counties and the mid size counties.
* Many rural areas are marginalized with lack of some types of service providers.
* Educational activities are the state's strong point, as most sites offer the required services.

* Staffing is somewhat inconsistently spread across the state, with the Indianapolis area approaching the goals and expectations identified in the CORN standards.
* A cost-benefit study may be necessary to determine the most effective and efficient model to organize and improve access in more remote areas of the state.
* Incentive programs may be necessary to attract new genetics counselors to the state and to enhance more equitable distribution of their services.
* A crucial resource available to existing practitioners is the education program "Genetics and Your Practice." This resource may need broader marketing to increase knowledge of its availability.

REFERENCES

Khoury M & Burke W (editors, et al). Genetics and Public Health in the 21st Century: Using Genetic Information to Improve Health and Prevent Disease. Oxford University Press: 2000. Chapter 5: The Multidisciplinary Nature of Public Health Genetics in Education. Pages 83-103.

TABLES

Geographical Location: NORTH		**Table 3.1: Services Offered by Location and Network** Type of Clinic: Primary or Satellite/Outreach Clinic Site & the Services Offered are indicated by								
Network Name/ Location	**Clinic Name / Location**	**Primary Clinic Site**	**Satellite/ Outreach Clinic Site**	**Genetic Evaluation & Counseling**	**Prenatal Diagnosis & Counseling**	**Cytogenetic Analysis**	**Ultrasound**	**Genetics Management Of High Risk Pregnancies**	**Pre-Conceptual Counseling**	**Testing for Inherited Cancers**
Gary	Gary									
Ft. Wayne	Ft. Wayne									
Ft. Wayne	South Bend									
IU/ Clarian/ Riley	South Bend									
Gary	South Bend									
Gary	Lake County									
Gary	Laporte									

Geographical Location: CENTRAL		Table 3.1: Services Offered by Location and Network (Continued) Type of Clinic: Primary or Satellite/Outreach Clinic Site & the Services Offered are indicated by								
Network Name/ Location	**Clinic Name / Location**	**Primary Clinic Site**	**Satellite/ Outreach Clinic Site**	**Genetic Evaluation & Counseling**	**Prenatal Diagnosis & Counseling**	**Cytogenetic Analysis**	**Ultrasound**	**Genetics Management Of High Risk Pregnancies**	**Pre-Conceptual Counseling**	**Testing for Inherited Cancers**
St. Vincent's/ Indianapolis	Indianapolis									
Center for Prenatal Diagnosis/ Indianapolis	Indianapolis									
IU/ Clarian/ Riley	Indianapolis									
Center for Prenatal Diagnosis/ Indianapolis	Lafayette									
Ft. Wayne	Muncie									
Community Hospitals/ Indianapolis	Indianapolis									
Indiana Center for Prenatal Diagnosis/ Indianapolis	Indianapolis									

Geographical Location: SOUTH		Table 3.1: Services Offered by Location and Network (Continued) Type of Clinic: Primary or Satellite/Outreach Clinic Site & the Services Offered are indicated by □								
Network Name/ Location	**Clinic Name/ Location**	**Primary Clinic Site**	**Satellite/ Outreach Clinic Site**	**Genetic Evaluation & Counseling**	**Prenatal Diagnosis & Counseling**	**Cytogenetic Analysis**	**Ultrasound**	**Genetics Management Of High Risk Pregnancies**	**Pre-Conceptual Counseling**	**Testing for Inherited Cancers**
Aegis/ Bloomington	Bloomington	□		□	□	□	□	□	□	□
IU/ Clarian/ Riley	Bloomington Hospital		□	□	□					
Center for Prenatal Diagnosis/ Indianapolis	Evansville		□	□	□		□	□		
IU/Clarian/Riley	Terre Haute		□	□	□	□	□	□	□	□
IU/Clarian/Riley	Evansville		□	□	□	□	□	□	□	□
IU/Clarian/Riley	Madison		□	□	□	□	□	□	□	□
Aegis/ Bloomington	Bedford		□	□	□	□	□	□	□	□
Aegis/ Bloomington	Linton		□	□	□	□	□	□	□	□
Clarksville	Clarksville	□		□	□	□	□	□	□	□
Clarksville	Corydon		□	□					□	
Clarksville	Jeffersonville		□	□				□	□	
St,. Mary's Regional Genetic Services Center	Evansville	□		□	□		□	□	□	
St,. Mary's Regional Genetic Services Center	Center for Prenatal Diagnosis/ Evansville		□	□	□		□	□	□	

Table 3.2: Genetic Specialty Staff in the State of Indiana by Network and Location

Personnel from the Primary Clinic of the Network staffing the Satellite/Outreach Sites, i.e., Shared Resources indicated by *****

Geographic Location: NORTH	**LOCATION/** Primary Clinic = **P** Satellite/Outreach Clinic = **S**	**Clinical Geneticist**	**Ph.D. Geneticist**	**Genetic Counselor**	**Cyto-Geneticist**	**Bio-Geneticist**	**Cytogenetic Technician**	**Genetic Nurse**	**Adv Nurse Practitioner**	**OB/GYN/ Peri-natologist**	**Diet Specialist**
Gary	Gary/**P**	1	0	0	1	0	1	0	0	0	0
Ft. Wayne	Ft. Wayne/**P**	1	0	1	1	0	7	0	1	0	0
Ft. Wayne	South Bend/**S**	1*	0	1*	1*	0	7*	0	1*	0	0
Gary	South Bend/**S**	1*	0	0	0	0	0	0	0	0	0
Gary	Lake County/**S**	1*	0	0	0	0	0	0	0	1	0
Gary	Laporte/**S**	1*	0	0	0	0	0	0	0	0	0
IU/Clarian/Riley	South Bend/**S**	3*	0	5*	2*	2*	13*	0	0	1*	0

Table 3.2: Genetic Specialty Staff in the State of Indiana by Network and Location (Continued)
Personnel from the Primary Clinic of the Network staffing the Satellite/Outreach Sites, i.e., Shared Resources indicated by *****

Geographic Location: CENTRAL	**LOCATION/** Primary Clinic = **P** Satellite/Outreach Clinic = **S**	**Clinical Geneticist**	**Ph.D. Geneticist**	**Genetic Counselor**	**Cyto-Geneticist**	**Bio-Geneticist**	**Cytogenetic Technician**	**Genetic Nurse**	**Advance Nurse Practitioner**	**OB/GYN/ Peri-natologist**	**Diet Specialist**
St. Vincent's/ Indianapolis	Indianapolis/ **P**	1	0	4	0	0	0	0	0	4	2
Center for Prenatal Diagnosis/ Indianapolis	Indianapolis/ **P**	0	0	2	0	0	0	0	0	1	0
IU/Clarian/Riley	Indianapolis/**P**	5	2	8	2	2	13	0	0	4	2
Center for Prenatal Diagnosis/ Indianapolis	Lafayette/ **S**	0	0	2*	0	0	0	0	0	1*	0
Ft. Wayne	Muncie/**S**	1*	0	1*	1*	0	7*	0	1*	0	0
Community Hospitals/ Indianapolis	Indianapolis/ **P**	1	0	0	0	1	2	0	1	1	2
Indiana Center for Prenatal Diagnosis/ Indianapolis	Indianapolis/ **P**	1	0	0	0	0	0	1	0	0	0

Table 3.2: Genetic Specialty Staff in the State of Indiana by Network and Location (Continued)
Personnel from the Primary Clinic of the Network staffing the Satellite/Outreach Sites, i.e., Shared Resources indicated by *****

Geographic Location: SOUTH	**LOCATION/** Primary Clinic = **P** Satellite/Outreach Clinic = **S**	**Clinical Geneticist**	**Ph.D. Geneticist**	**Genetic Counselor**	**Cyto-Geneticist**	**Bio-Geneticist**	**Cytogenetic Technician**	**Genetic Nurse**	**Advanced Nurse Practitioner**	**OB/GYN/ Peri-natologist**	**Diet Specialist**
Aegis/ Bloomington	Bloomington/ **P**	0	0	1	0	0	0	0	0	1	1
IU/Clarian/Riley	Bloomington Hospital/**S**	7*	2*	5*	2*	2*	13*	0	0	1*	2*
Center for Prenatal Diagnosis/ Indianapolis	Evansville/**S**	0	0	2*	0	0	0	0	0	1*	0
IU/Clarian/Riley	Terre Haute/**S**	2*	2*	2*	2*	2*	13*	0	0	1*	2*
IU/Clarian/Riley	Madison/**S**	3*	2*	5*	2*	2*	13*	0	0	1*	2*
IU/Clarian/Riley	Evansville/**S**	3*	2*	5*	2*	2*	13*	0	0	1*	2*
Aegis/Bloomington	Bedford/**S**	0	0	1*	0	0	0	0	0	1*	1*
Aegis/Bloomington	Linton/**S**	0	0	1*	0	0	0	0	0	1*	1*
Clarksville	Clarksville/**P**	3	0	2	0	0	0	0	0	0	0
Clarksville	Corydon/**S**	3*	0	2*	0	0	0	0	0	0	0
Clarksville	Jeffersonville/**S**	3*	0	2*	0	0	0	0	0	0	0
St. Mary's Regional Genetic Services Center	Evansville/**P**	1	0	1	0	0	0	0	0	1	2
St. Mary's Regional Genetic Services Center	Center for Prenatal Diagnosis/ Evansville/**S**	0	0	1*	0	0	0	0	0	1	0

Table 3.3: Clinical and Educational Genetic Services in the State of Indiana by Network and Location

Clinical & Educational Services Offered indicated by □

Geographic Location: NORTH	**LOCATION/** Primary Clinic = **P** Satellite/ Outreach Clinic = **S**	**Clinical Genetic Services**					**Educational Genetic Services**				**Utilization & Funding**	
		Screenings				**Laboratory**					**Utilization**	**Funding**
		Adult	Child	Newborn	Prenatal	**B** = Biochemical Genetics **C** = Cytogenetic **M** = Molecular Genetics	Family Screening	Folic Acid	Terato-genetic	Other	Number Served/ Year	Source of Funding : □ Private □ Public
Gary	Gary/**P**			□	□	**C**		□	□		950	Private &Public
Ft. Wayne	Ft. Wayne/**P**	□	□	□	□	**C**	□	□	□		1,200	Private & Public
Ft. Wayne	South Bend/**S**	□	□	□	□	**C**	□	□	□		Unknown	Private & Public
Gary	South Bend/**S**			□	□	**C**		□	□		Unknown	Private &Public
Gary	Lake County/**S**			□	□	**C**		□	□		Unknown	Private &Public
Gary	Laporte/**S**			□	□	**C**		□	□		Unknown	Private &Public
IU/ Clarian/ Riley	South Bend/**S**					**C**	□	□	□	MD: Education & Training	Unknown	Private & Public

Table 3.3: Clinical and Educational Genetic Services in the State of Indiana by Network and Location (Continued)
Clinical & Educational Services Offered indicated by □

Geographic Location: CENTRAL	**LOCATION/** Primary Clinic = **P** Satellite/ Outreach Clinic = **S**	**Clinical Genetic Services**					**Educational Genetic Services**				**Utilization** & **Funding**	
		Screenings				**Laboratory**					**Utilization**	**Funding**
		Adult	Child	Newborn	Prenatal	**B** = Biochemical Genetics **C** = Cytogenetic **M** = Molecular Genetics	Family Screening	Folic Acid	Terato-genetic	Other	Number Served/ Year	Source of Funding : □ Private □ Public
St. Vincent's/ Indianapolis	Indianapolis/ **P**	□	□	□	□	**None**	□	□	□	MD: Education & Training	1,100	Private &Public
Center for Prenatal Diagnosis/ Indianapolis	Indianapolis/ **P**			□	□	**None**		□	□		5,100	Private
IU/Clarian/ Riley	Indianapolis/ **P**					**C**	□	□	□	MD: Education & Training	12,000	Private & Public
Center for Prenatal Diagnosis/ Indianapolis	Lafayette/ **S**			□	□	**None**		□	□		Unknown	Private
Ft. Wayne	Muncie/**S**	□	□	□	□	**C**	□	□	□		Unknown	Private & Public
Community Hospitals/ Indianapolis	Indianapolis/ **P**	□	□	□	□	**C**	□	□	□		Unknown	Private
Indiana Center for Prenatal Diagnosis/ Indianapolis	Indianapolis/ **P**			□	□	**None**		□	□		Unknown	Private

Table 3.3: Clinical and Educational Genetic Services in the State of Indiana by Network and Location (Continued)
Clinical & Educational Services Offered indicated by

Geographic Location: SOUTH	LOCATION/ Primary Clinic = **P** Satellite/ Outreach Clinic = **S**	Clinical Genetic Services					Educational Genetic Services				Utilization & Funding	
		Screenings				Laboratory					Utilization	Funding
		Adult	Child	Newborn	Prenatal	**B** = Biochemical Genetics **C** = Cytogenetic **M** = Molecular Genetics	Family Screening	Folic Acid	Terato-genetic	Other	Number Served/ Year	Source of Funding : Private Public
Aegis/ Bloomington	Bloomington/ **P**					**None**					1,300	Private &Public
IU/Clarian/ Riley	Bloomington Hospital/**S**					**C**				MD: Education & Training	Unknown	Private & Public
Center for Prenatal Diagnosis/ Indianapolis	Evansville/**S**					**None**					Unknown	Private
IU/Clarian/ Riley	Terre Haute/**S**					**C**				MD: Education & Training	Unknown	Private & Public
IU/Clarian/ Riley	Madison/**S**					**C**				MD: Education & Training	Unknown	Private & Public
Aegis/ Bloomington	Bedford/**S**					**None**					Unknown	Private &Public
Aegis/ Bloomington	Linton/**S**					**None**					Unknown	Private &Public
Clarksville	Clarksville/**P**					**None**					700	Private &Public
Clarksville	Corydon/**S**					**None**					Unknown	Private &Public
Clarksville	Jeffersonville /**S**					**None**					Unknown	Private &Public

Table 3.3: Clinical and Educational Genetic Services in the State of Indiana by Network and Location (Continued)
Clinical & Educational Services Offered indicated by

Geographic Location: SOUTH	**LOCATION/** Primary Clinic = **P** Satellite/ Outreach Clinic = **S**	**Clinical Genetic Services**					**Educational Genetic Services**				**Utilization** & **Funding**	
		Screenings				**Laboratory**					**Utilization**	**Funding**
		Adult	Child	Newborn	Prenatal	**B** = Biochemical Genetics **C** = Cytogenetic **M** = Molecular Genetics	Family Screening	Folic Acid	Terato-genetic	Other	Number Served/ Year	Source of Funding : Private Public
IU/Clarian/ Riley	Evansville /**S**					**C**				MD: Education & Training	Unknown	Private & Public
St,. Mary's Regional Genetic Services Center	Evansville/**P**					**None**					300	Private & Public
St,. Mary's Regional Genetic Services Center	Center for Prenatal Diagnosis/ Evansville/**S**					**None**					Unknown	Private & Public

Table 3.4: All First Steps Service Providers

Grand Total	3149
Speech Pathologist	686
Physical Therapist	535
Occupational Therapist	481
Developmental/Educational Specialist	378
Service Coordinator	304
Audiologist	109
Physical Therapy Assistant	88
Service Coordinator Associate	79
Interpreter	61
Occupational Therapy Assistant	56
Common Transport (Ambulatory)	55
Social Worker (MSW)	50
Psychologist	42
Registered Dietitian	42
Registered Nurse	40
Developmental/Educational Associate	27
Otolaryngologist (ENT physician)	20
Speech Pathologist/Audiologist Aide	15
Pediatrician	13
Marriage and Family Therapist	11
Speech/Hearing Therapist	9
Vision Specialist	9
Alternative Therapy	8
Licensed Practical Nurse	8
Rehabilitation Services	7
Common Transport (Non-Ambulatory)	5
Orientation/Mobility Specialist	2
Ambulance	1
Bus	1
Developmental/Educational Assistant	1
General Surgeon	1
Intermediate Care Facility/Mental Retardation	1
Ophthalmologist	1
Orthopedic Surgeon	1
Psychiatrist	1
Speech Hearing Therapist	1

Table 3.5: Classes of First Steps Service Providers

Speech and Hearing	
	Audiologist
	Speech Hearing Therapist
	Speech Pathologist/Audiologist Aide
	Speech Pathologist
	Speech/Hearing Therapist
Alternative Therapy	
	Alternative Therapy
Developmental Services	
	Developmental/Educational Associate
	Developmental/Educational Assistant
	Developmental/Educational Specialist
Transportation	
	Ambulance
	Bus
	Common Transport (Non-Ambulatory)
	Common Transport (Ambulatory)
Medical Physicians	
	General Surgeon
	Intermediate Care Facility/Mental Retardation
	Ophthalmologist
	Orientation/Mobility Specialist
	Orthopedic Surgeon
	Otolaryngologist (ENT physician)
	Pediatrician
Interpreter	
	Interpreter
Nurses	
	Licensed Practical Nurse (LPN)
	Registered Nurse (RN)
Allied Health	
	Occupational Therapist
	Occupational Therapy Assistant
	Physical Therapy Assistant
	Physical Therapist
	Rehabilitation Services
Counseling Services	
	Marriage and Family Therapist
	Psychiatrist
	Psychologist
	Social Worker (MSW)
Registered Dietitian	
	Registered Dietitian
Service Coordination	
	Service Coordinator
	Service Coordinator Associate
Vision	
	Vision Specialist

Table 3.6: Distribution of Physicians by County and Specialty

County	Type	Family	GP	Internal	OB/GYN	Peds	Total	%	Cum %
Total		1381	260	575	362	387	2965	100%	N/A
Marion	Metro	208	24	157	90	105	584	19.7%	19.7%
Lake	Metro	109	32	57	40	42	280	9.4%	29.1%
Saint Joseph	Metro	82	11	18	19	14	144	4.9%	34.0%
Allen	Metro	80	8	15	16	19	138	4.7%	38.7%
Vanderburgh	Metro	57	5	30	17	20	129	4.4%	43.0%
Hamilton	Metro	40	1	18	9	13	81	2.7%	45.7%
Elkhart	Metro	40	2	10	16	9	77	2.6%	48.3%
Tippecanoe	Metro	24	4	16	12	15	71	2.4%	50.7%
Vigo	Metro	30	5	11	10	10	66	2.2%	53.0%
Clark	Metro	27	6	14	8	8	63	2.1%	55.1%
Delaware	Metro	23	6	20	5	8	62	2.1%	57.2%
Madison	Metro	35	8	7	5	5	60	2.0%	59.2%
Monroe	Metro	21	11	6	12	5	55	1.9%	61.0%
Porter	Metro	23	4	10	6	8	51	1.7%	62.8%
LaPorte	Metro	18	7	11	5	7	48	1.6%	64.4%
Howard	Metro	26	0	11	5	5	47	1.6%	66.0%
Bartholomew	Metro	20	1	8	7	10	46	1.6%	67.5%
Johnson	Metro	22	2	7	3	10	44	1.5%	69.0%
Hendricks	Metro	27	2	3	5	4	41	1.4%	70.4%
Floyd	Metro	16	4	3	4	7	34	1.1%	71.5%
Wayne	Metro	14	2	13	3	1	33	1.1%	72.6%
Boone	Metro	11	3	8	2	5	29	1.0%	73.6%
Grant	Metro	11	6	4	4	3	28	0.9%	74.6%
Kosciusko	Metro	18	1	2	2	2	25	0.8%	75.4%
Hancock	Metro	15	1	3	4	1	24	0.8%	76.2%
Henry	Metro	12	3	5	2	2	24	0.8%	77.0%
Dearborn	Metro	10	3	4	1	5	23	0.8%	77.8%
Knox	Metro	7	1	8	2	3	21	0.7%	78.5%
Warrick	Metro	11	4	2	2	2	21	0.7%	79.2%
Huntington	Metro	11	3	2	2	2	20	0.7%	79.9%
Montgomery	Metro	12	0	4	1	2	19	0.6%	80.5%
Wabash	Metro	15	1	1	2	0	19	0.6%	81.2%
Cass	Metro	8	6	3	0	1	18	0.6%	81.8%
DeKalb	Metro	13	3	1	1	0	18	0.6%	82.4%
Morgan	Metro	5	2	5	2	2	16	0.5%	82.9%
Jefferson	Metro	6	3	3	1	2	15	0.5%	83.4%
Miami	Metro	7	1	2	3	1	14	0.5%	83.9%
Fayette	Metro	5	2	3	2	1	13	0.4%	84.4%
Harrison	Metro	8	1	2	1	1	13	0.4%	84.8%
Jackson	Metro	7	3	1	1	0	12	0.4%	85.2%
Clinton	Metro	5	1	3	2	0	11	0.4%	85.6%
Shelby	Metro	5	1	3	1	0	10	0.3%	85.9%
Daviess	Metro	5	1	0	1	2	9	0.3%	86.2%
Clay	Metro	4	2	0	0	1	7	0.2%	86.4%
Posey	Metro	5	1	1	0	0	7	0.2%	86.7%
Whitley	Metro	4	1	1	1	0	7	0.2%	86.9%
Tipton	Metro	5	0	0	1	0	6	0.2%	87.1%

Table 3.6: Distribution of Physicians by County and Specialty (Continued)

County	Type	Family	GP	Internal	OB/GYN	Peds	Total	%	Cum %
Marshall	Rural	20	4	2	1	3	30	1.0%	88.1%
Wells	Rural	10	1	5	4	5	25	0.8%	89.0%
Dubois	Rural	10	0	5	4	2	21	0.7%	89.7%
Gibson	Rural	11	0	5	2	1	19	0.6%	90.3%
Ripley	Rural	4	4	3	2	4	17	0.6%	90.9%
Steuben	Rural	12	2	0	0	0	14	0.5%	91.4%
Noble	Rural	10	3	0	0	0	13	0.4%	91.8%
Putnam	Rural	9	3	0	1	0	13	0.4%	92.2%
Fulton	Rural	5	3	1	0	2	11	0.4%	92.6%
Jasper	Rural	11	0	0	0	0	11	0.4%	93.0%
Jennings	Rural	4	3	4	0	0	11	0.4%	93.4%
Washington	Rural	5	2	3	0	1	11	0.4%	93.7%
Decatur	Rural	2	2	3	2	1	10	0.3%	94.1%
Adams	Rural	7	2	0	0	0	9	0.3%	94.4%
Sullivan	Rural	4	1	4	0	0	9	0.3%	94.7%
Jay	Rural	8	0	0	0	0	8	0.3%	94.9%
LaGrange	Rural	8	0	0	0	0	8	0.3%	95.2%
White	Rural	6	2	0	0	0	8	0.3%	95.5%
Carroll	Rural	7	0	0	0	0	7	0.2%	95.7%
Greene	Rural	4	1	1	1	0	7	0.2%	96.0%
Orange	Rural	7	0	0	0	0	7	0.2%	96.2%
Scott	Rural	6	0	1	0	0	7	0.2%	96.4%
Blackford	Rural	2	4	0	0	0	6	0.2%	96.6%
Parke	Rural	4	2	0	0	0	6	0.2%	96.8%
Randolph	Rural	1	1	2	1	1	6	0.2%	97.0%
Rush	Rural	3	2	1	0	0	6	0.2%	97.2%
Fountain	Rural	2	1	1	0	1	5	0.2%	97.4%
Pulaski	Rural	3	1	0	1	0	5	0.2%	97.6%
Starke	Rural	3	1	1	0	0	5	0.2%	97.7%
Vermillion	Rural	2	2	1	0	0	5	0.2%	97.9%
Benton	Rural	4	0	0	0	0	4	0.1%	98.0%
Perry	Rural	3	1	0	0	0	4	0.1%	98.2%
Pike	Rural	3	0	1	0	0	4	0.1%	98.3%
Franklin	Rural	1	0	2	0	0	3	0.1%	98.4%
Martin	Rural	0	1	2	0	0	3	0.1%	98.5%
Owen	Rural	1	1	1	0	0	3	0.1%	98.6%
Switzerland	Rural	1	2	0	0	0	3	0.1%	98.7%
Warren	Rural	1	2	0	0	0	3	0.1%	98.8%
Brown	Rural	1	1	0	0	0	2	0.1%	98.9%
Crawford	Rural	1	0	1	0	0	2	0.1%	99.0%
Newton	Rural	1	0	1	0	0	2	0.1%	99.0%
Ohio	Rural	0	0	1	0	0	1	0.0%	99.1%
Spencer	Rural	0	1	0	0	0	1	0.0%	99.1%
Union	Rural	1	0	0	0	0	1	0.0%	99.1%

FIGURES

Figure 3.1: Institutional Criteria

Services Offered:

* Genetic Evaluation and Counseling
* Prenatal Diagnosis and Counseling
* Cytogenetic Analysis
* Ultrasonography
* Genetics Management of High Risk Pregnancies
* Pre-Conceptual Counseling
* Testing for Inherited Cancers

Staff at Hospital (number, etc.):

* Clinical Geneticist
* Ph.D. Medical Geneticist
* Genetic Counselor
* Clinical Cytogeneticist
* Clinical Biochemical Geneticist
* Cytogenetic Technologist
* Genetic Nurse
* Advanced Practice Nurse/Geneticist
* Perinatologist/Obstetrician
* Dietician
* Other

Type of Educational Activities Offered:

* Type: Folic Acid Education
* Teratogenetic Education Information Systems, (educational, management, access, etc.)
* Other Services
* Number Served
* Funding
* Family Based Screenings

Types of (Population) Screening Offered at Each Institution:

* Prenatal Screening
* Newborn Screening
* Childhood Screening
* Adult Screening

Clinical Laboratory Services:

* Cytogenetics
* Biochemical Genetics
* Molecular Genetics

Figure 3.2: Service Networks in Indiana; Part I

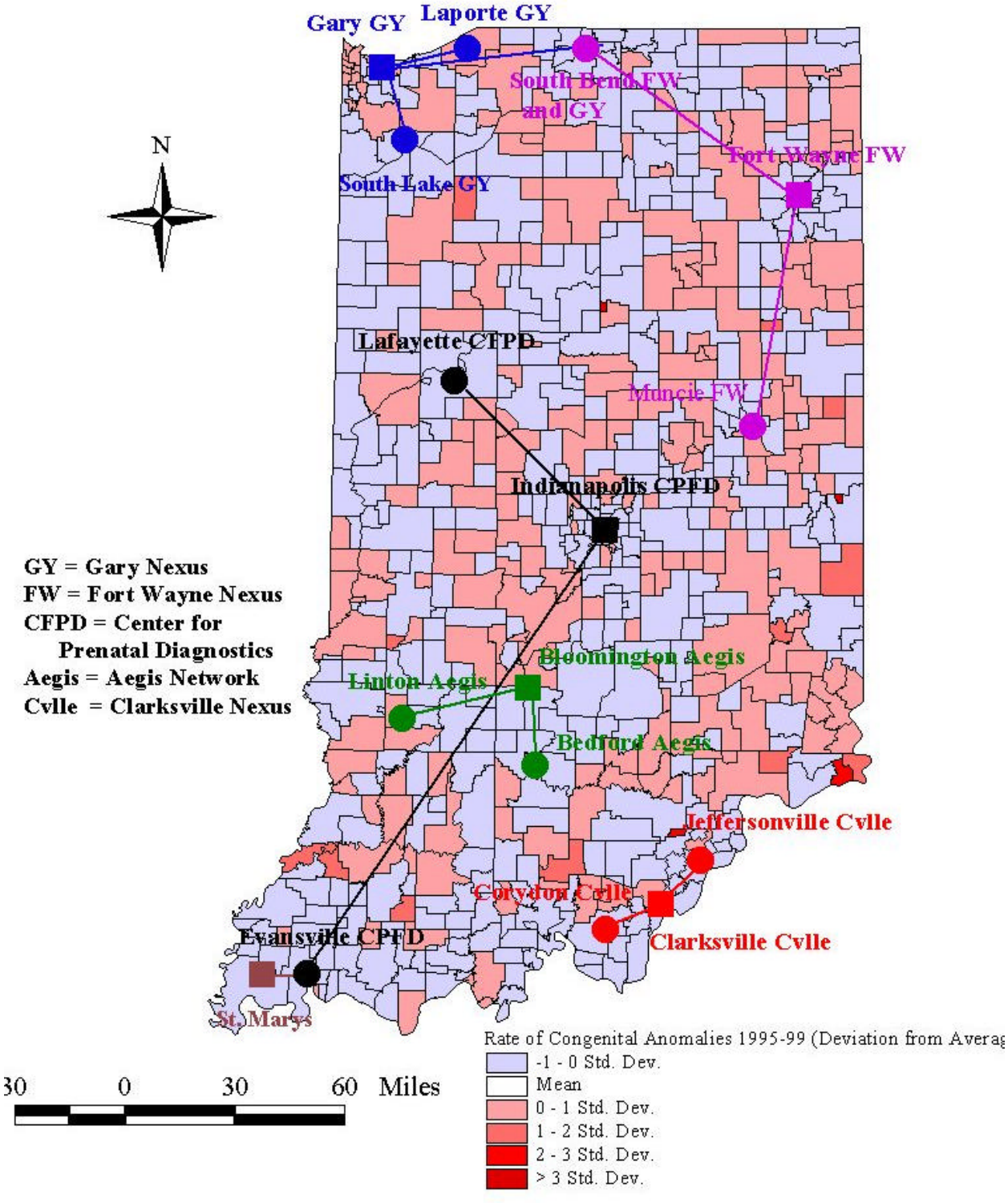
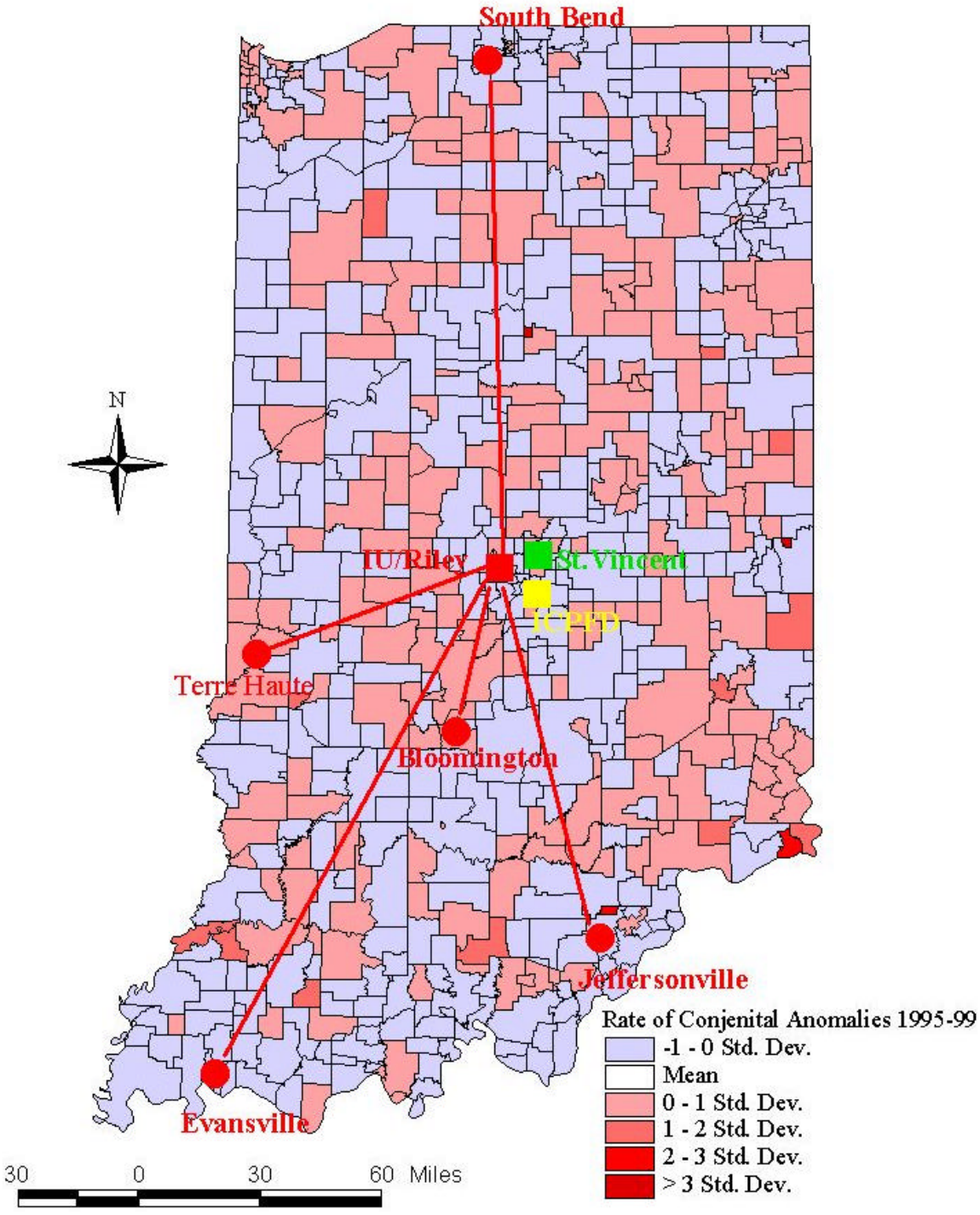


Figure 3.3: Service Networks in Indiana; Part II
IU/Riley & Indianapolis Non-NetworkSites



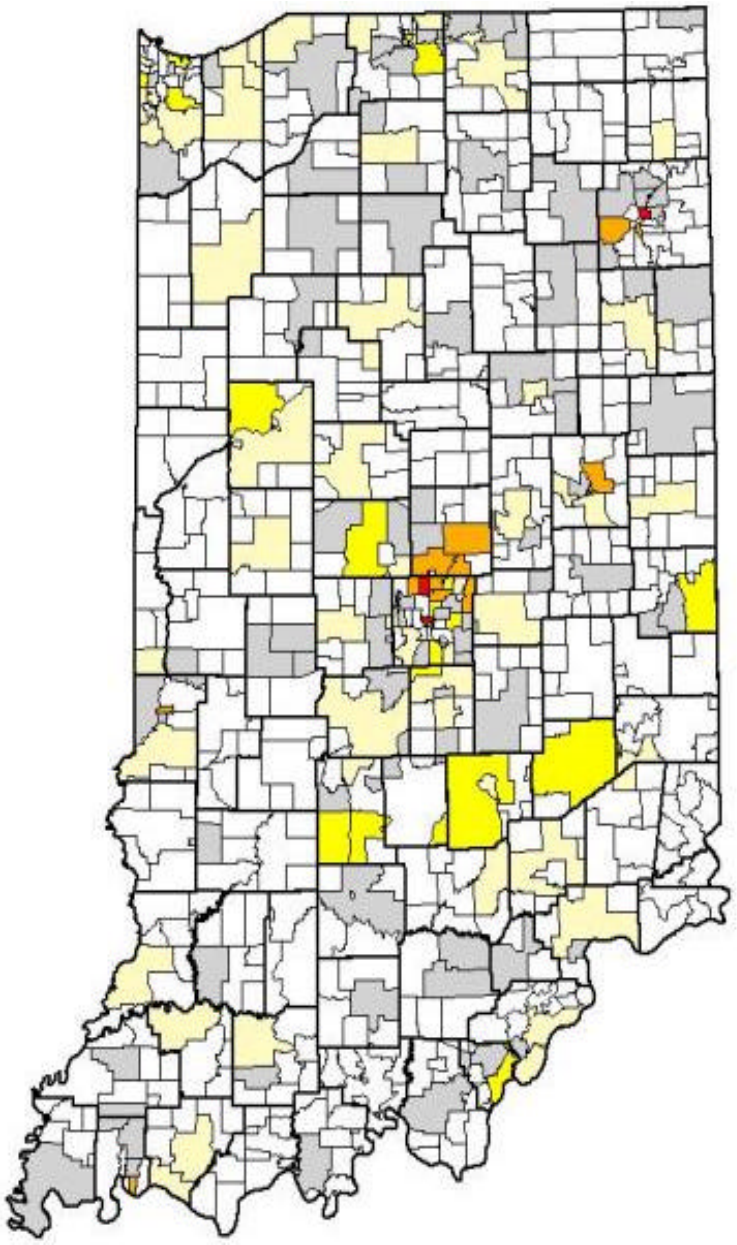
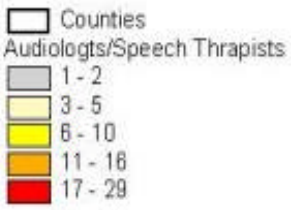


Figure 3.4:
Audiologist and Speech Therapists

(By ZIP Code)



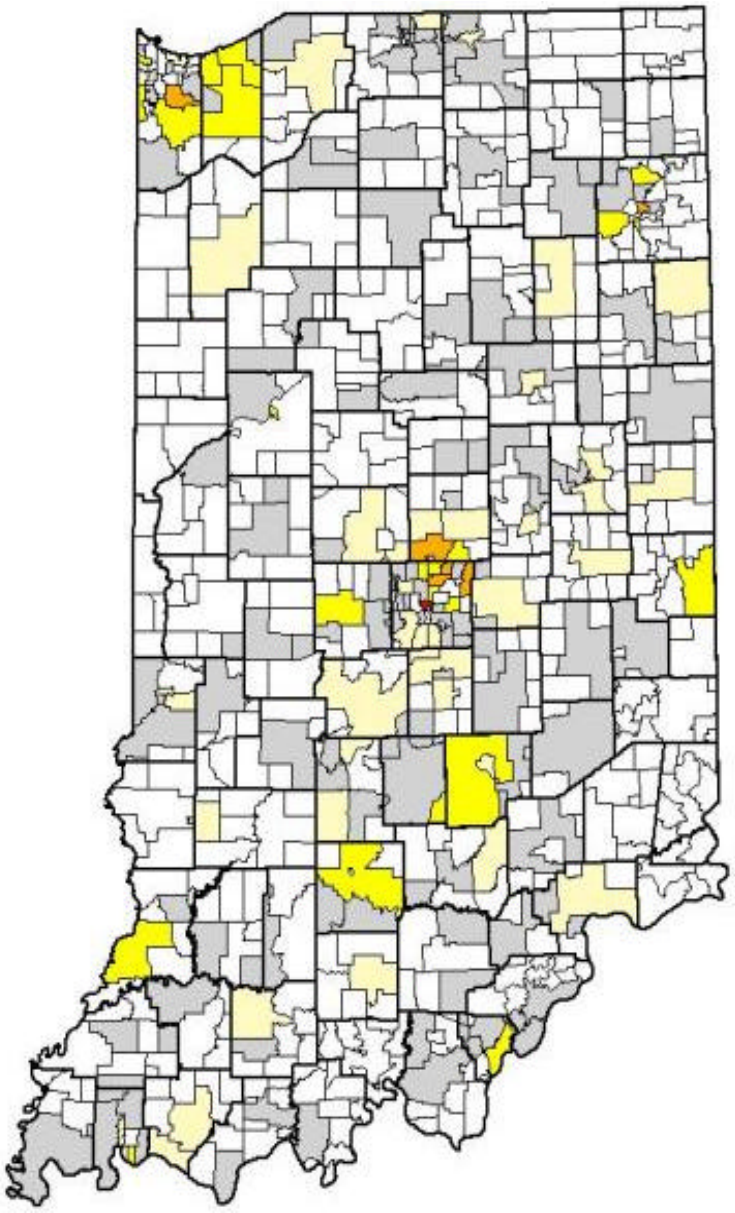


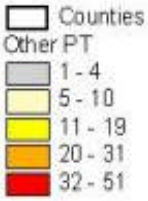
Figure 3.5:

Counselors,

Psychiatric &

Psychological Services

(By ZIP Code)



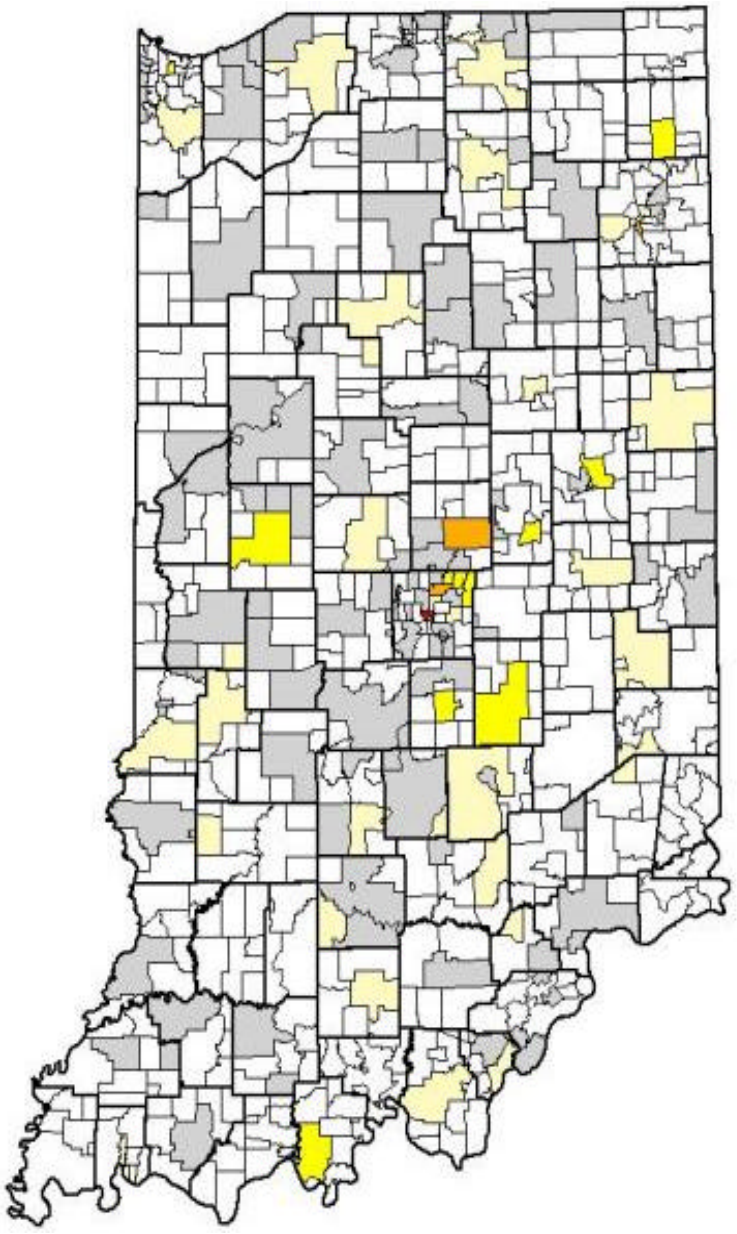


Figure 3.6:
Developmental Service Providers/Coordinators

(By ZIP Code)

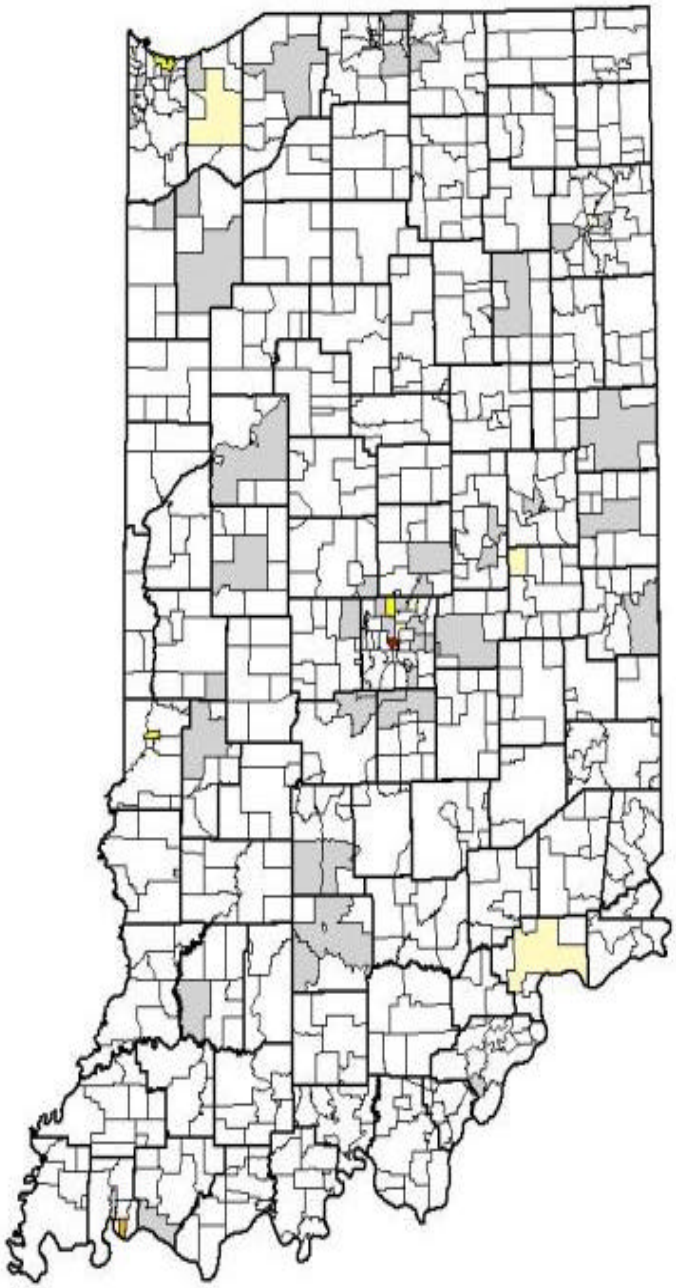
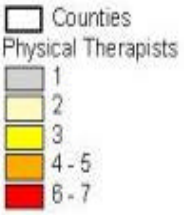


Figure 3.7:

Physical Therapists

(By ZIP Code)



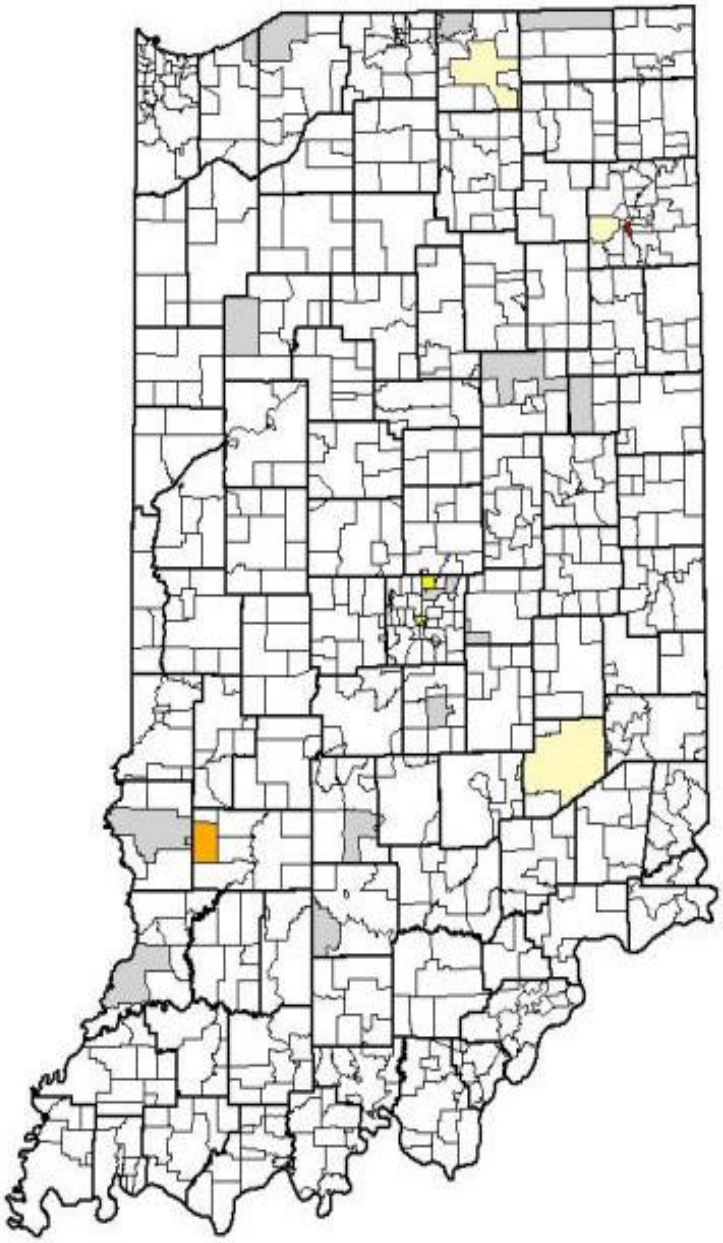
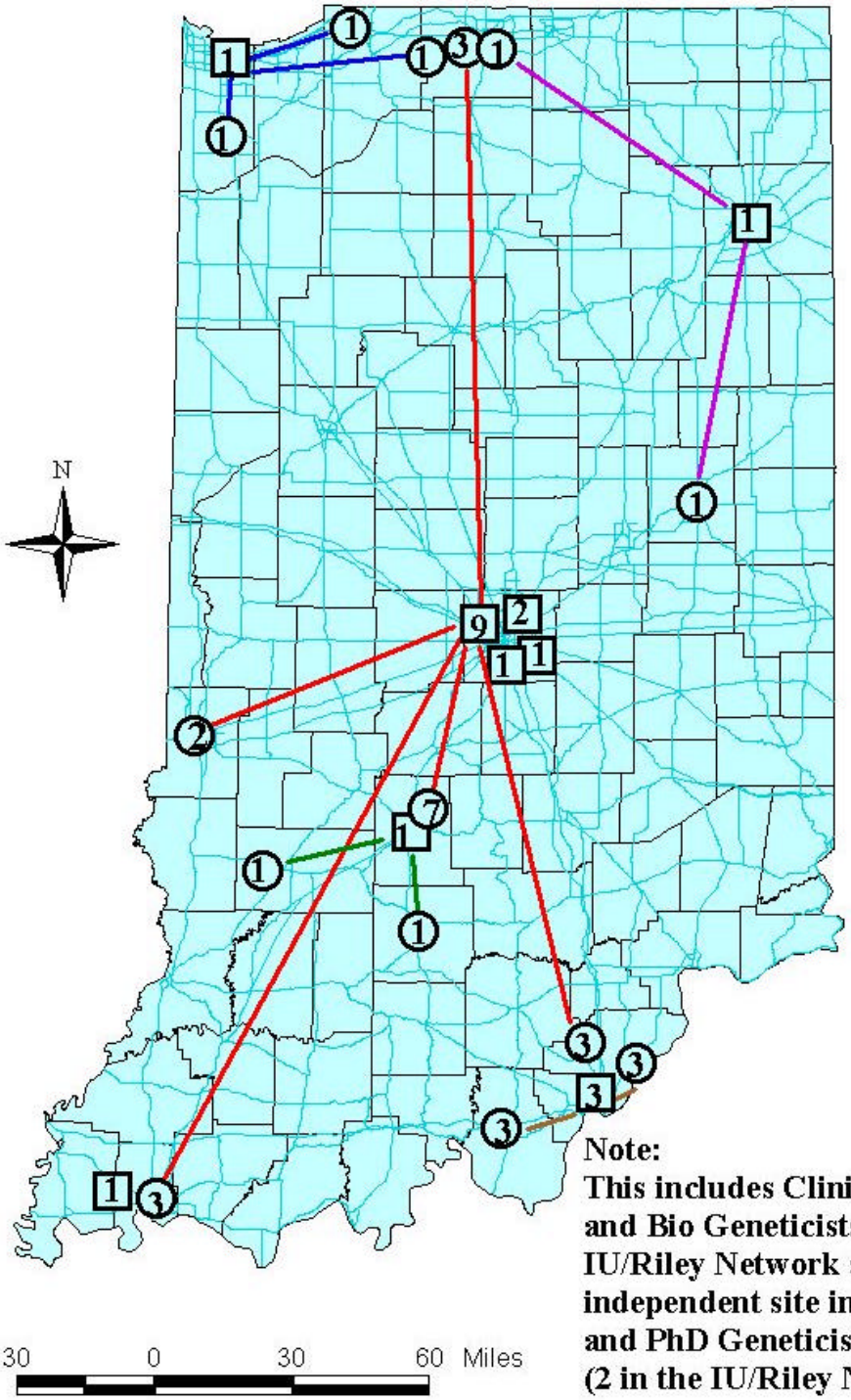


Figure 3.8:
Allied Medicine Providers-Nurses

(By ZIP Code)

Figure 3.9: Geneticists by Network



Note:
This includes Clinical Geneticists and Bio Geneticists (2 in the IU/Riley Network and 1 at an independent site in Indianapolis) and PhD Geneticists (2 in the IU/Riley Network)

Figure 3.10: Genetic Counselors by Network

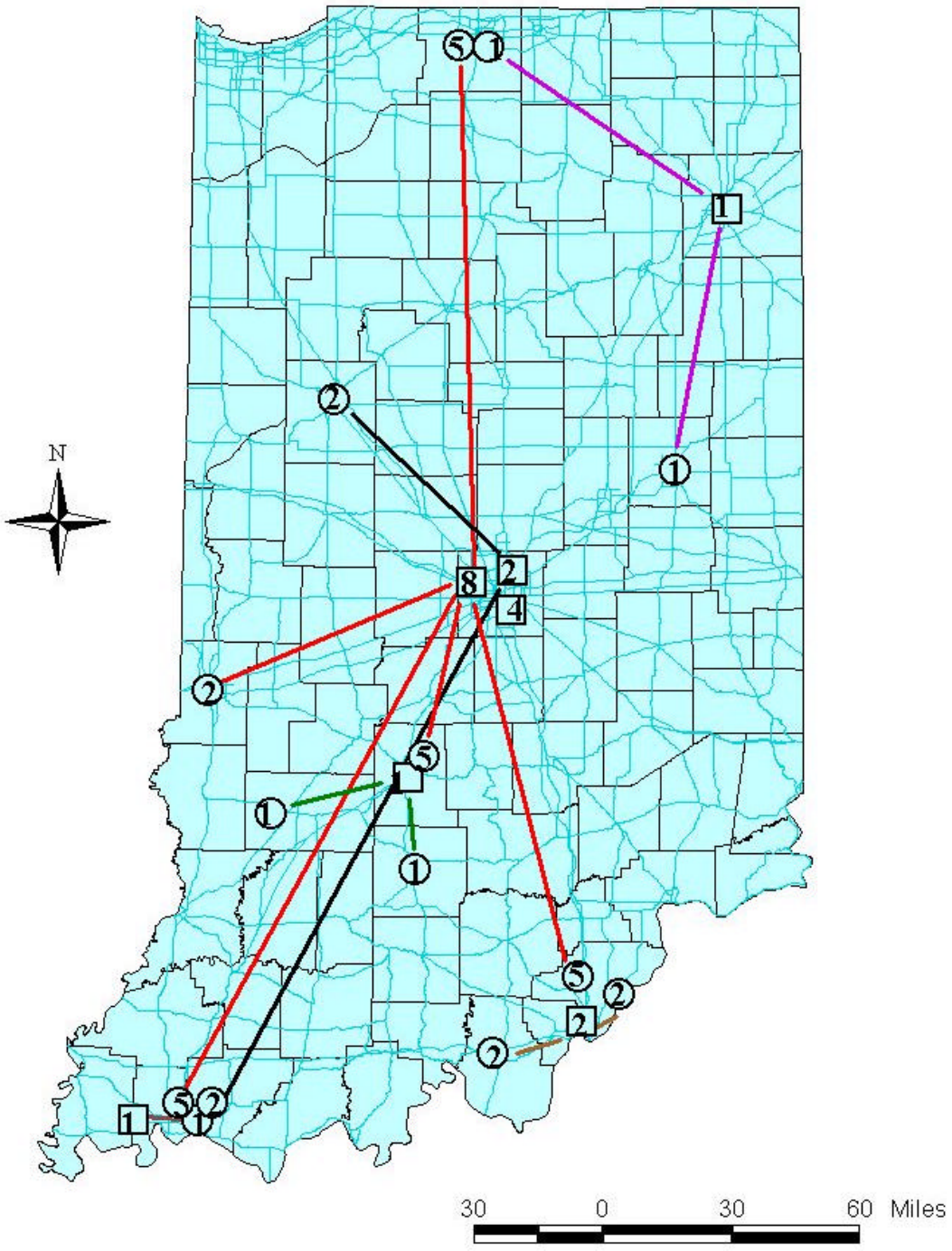
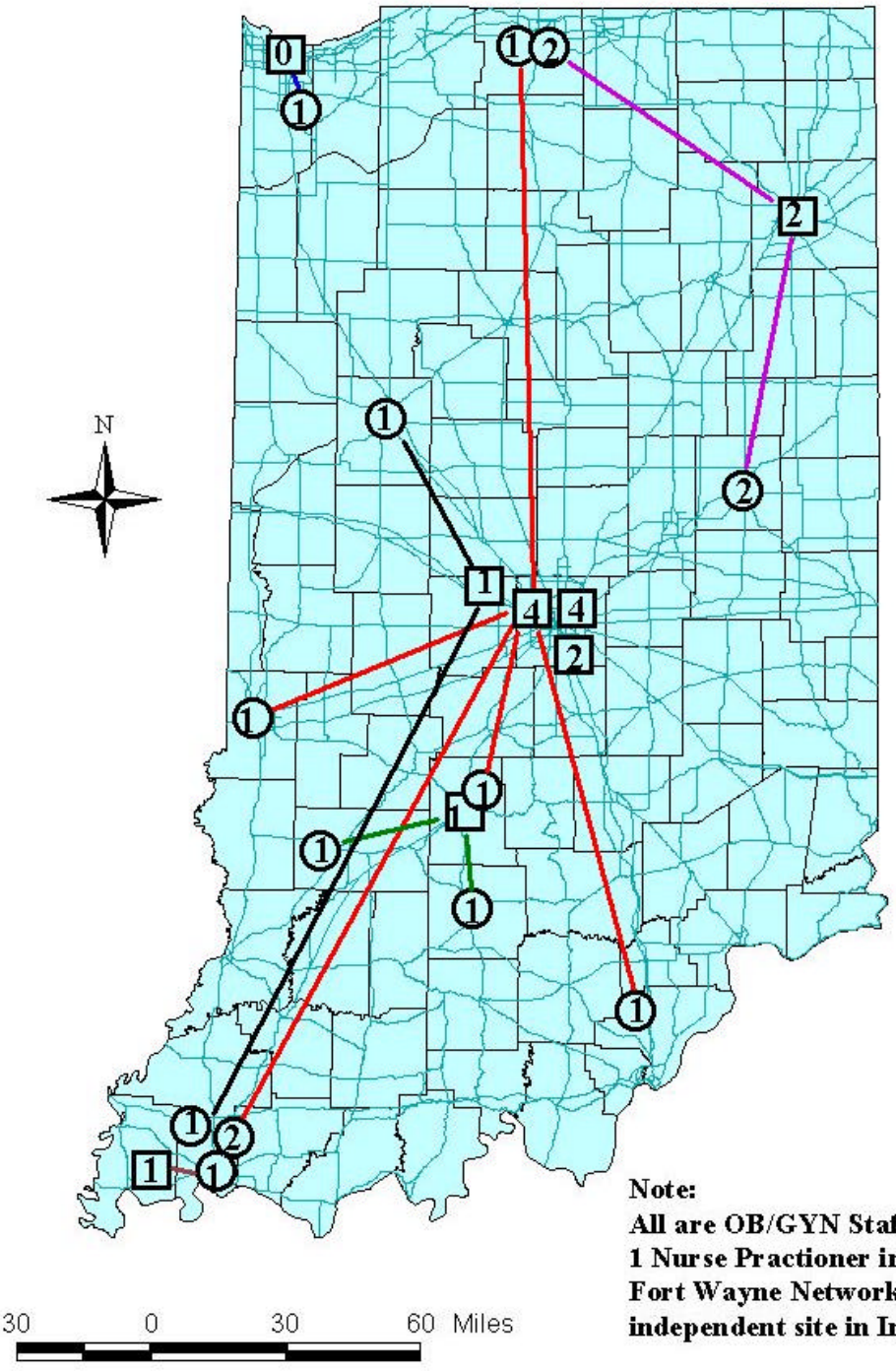


Figure 3.11: OB/GYN Staff and Nurse Practitioners With Genetic Specialty by Network



Note:
All are OB/GYN Staff except 1 Nurse Practioner in the Fort Wayne Network and 1 at an independent site in Indianapolis

4.0 PUBLIC AND PROFESSIONAL PERCEPTIONS AND CONCERNS

Sections 2.0 and 3.0 of this report focus on hard numbers necessary to identify need/demand for services and the capacity of current infrastructure to respond to that demand. However, public perceptions and understanding of the difficulties are also key factors in determining access and demand issues. This section details three different measures of public perceptions of need, problems and access. A key informant survey was conducted in May and June to determine perceptions of selected individuals who constituted knowledgeable service providers, family members and decision makers in the state. Information gathered from First Steps focus groups across the state was integrated into the needs assessment. And finally, a special survey directed toward physicians was completed in July and August.

4.1 Key Informant Interviews

4.1.1 Background

The qualitative approach to data collection is used to collect information not easily assessed in an objective, quantitative manner (Kruger, 1994). Although not easily gathered, qualitative data collected using planned methods and instruments may provide a fuller understanding of the context of a problem. Generally, qualitative data reveal attitudes, perceptions, and opinions of those included in the study (Kruger, 1994). Furthermore, key informants, those deemed experienced or those having in-depth knowledge in a subject, are an important component of qualitative research since these individuals are usually responsible for leading the vision or influencing the future of an issue or process.

Key informant interviews (targeted thoughts of leaders involved in the infrastructure of birth defects and genetic conditions services) were included in the needs assessment for the Genetics Planning Grant. The purpose of these interviews was to assess key informant opinions and perceptions of access, quality and availability of genetic services.

4.1.2 Methodology

The key informant interviews were conducted first to serve as a platform for the other components of the needs assessment process. The results of the interviews were used to develop the provider surveys and assess existing focus group data.

The interview instrument (Appendix D) was designed by faculty and staff at the Indiana University-Bowen Research Center (IUBRC) and reviewed by the Indiana State Department of Health Genetics Planning Grant Strategic Planning Committee. The development of the instrument consisted of three major planning phases: 1) conceptualization of the purpose and goals of the study, 2) development of an initial draft of the instrument, and 3) planning of logical and fluid question order. During the development of the initial draft, survey instruments designed by other states receiving Genetics Planning Grants were gathered and reviewed as a guide.

The final instrument for the key informant surveys consisted of ten questions with the flexibility for "probing" that could be included to clarify responses. A protocol (Appendix E) was written to assist interviewers in establishing interviewer-consistency and rapport. Information pertaining to interviewer neutrality was also included. Two scripts (Appendix F) were written to trigger the interviewer to complete all steps of the interview process. Script 1 was used at the time of the

initial phone call. The script included a formal introduction, description of the purpose of the study, assurance of confidentiality of responses, and scheduling of the interview appointments. Script 2 re-stated the description, purpose and once again assured confidentiality. A summary sheet was used during the interview process to record responses and document probing questions.

4.1.3 Data Collection Process and Coding

Telephone calls were made to key informants to schedule interviews. Script 1 was used to plan the interview. On the scheduled date, interviewers called the key informants and initiated the interviews using Script 2. Responses to interview and probing questions were documented on the summary sheet. During the interview, words, phrases and their context used by informants were considered when summarizing responses. The intensity, extensiveness, and specificity of responses were also used in the interpretation of the comments.

Questions were organized in a manner to promote an effective response flow from key informants. Common, uncommon and inconsistent responses were labeled and addressed more thoroughly following the interview. Interviewers also verified responses to assure accuracy. In preparation for analysis, interviewers underlined common words used throughout all of the interviews.

For the final analysis, emerging themes for each question and overall survey were identified. Professions were categorized into four main areas: medical geneticists; genetics-related professions such as genetics counselors; other medical professions (physicians dealing with the pediatric population); and related professions (administrative and auxiliary health services support). All questions were analyzed and reported by professional category using the cross-tabulation procedure in Statistical Package for the Social Sciences (SPSS) version10.0.

4.1.4 Results

Thirty-six key informants from a variety of disciplines were interviewed. Respondents were asked to provide additional key informants not currently on our list at the end of each interview. Any additional key informants were added to the initial call list provided by the state and targeted for interview.

The analysis of participants by professional category indicated fifteen (41.7%) of the key informants were in the related professions category, e.g., administrative and auxiliary health services support. Eleven (30.6%) of the participants were classified as genetics-related professionals. Of the remaining participants five (13.9%) were categorized as medical geneticists, and five (13.9%) were categorized as other medical professions (all included in this category were physicians).

At least sixty percent (60%) of all professions indicated the access to services for citizens with birth defects and genetics conditions was fair. Another fourteen percent (14%) thought access to services was good to excellent. Key informants were asked to identify barriers to access. Multiple responses were reported. The key informants indicated location (61%), cost reimbursement (56%), and lack of knowledge of the providers and consumers of services (47%) as the most significant barriers. However, when asked about quality of services, over half (53%)

indicated available services were excellent. Overall, eighty-six percent (86%) considered services good to excellent. Conversely, eighty-one percent (81%) of the key informants reported there are not enough physicians, clinical providers and genetics counselors to meet the service needs of citizens with birth defects and genetics conditions.

Of the key informants participating, fifty percent (50%) rated the adequacy of medical training as fair in relationship to birth defects and genetic conditions. Only seventeen percent (17%) of the total participants thought the physicians and health care providers were trained to identify birth defects and genetic conditions following birth. Additionally, fourteen percent (14%) considered the information they have, in relationship to birth defects and genetic conditions, enough to provide appropriate referrals.

Key informants were asked three questions pertaining to the improved quality of information and services. Seventy-two percent (72%) favored written and verbal communication (i.e., brochures and pamphlets, follow-up telephone calls, and follow-up letters) to convey information to the patient. Suggestions for improvements and expansion of the current health care system focused on reimbursement and financial support. Key informants thought grants, both state and federal money to support services and programs and expansion of services covered by both private and public payers would, be very helpful. Expansion of the educational programs for providers and consumers would also be advantageous in the expansion of the health care system for citizens with birth defects and genetics conditions. It was also noted that financial support was needed at the client level.

4.1.5 Themes from Key Informants

Three prevailing themes occurred in these data. The first theme dealt with the issue of service disbursement throughout the State of Indiana. Key informants noted birth defects and genetics conditions services were not distributed equally throughout the State. It was the opinion of the key informants that services were limited in rural areas compared to more densely populated areas and often consumers must travel long distance to receive services. The second theme that appeared was focused on the education of the physician. Key informants had some difficulty answering questions related to physician and health care provider training. Most thought the primary care provider needed further training in detection, identification, treatment, and appropriate referral of patients with birth defects and genetics conditions. However, specialists were viewed as educated and informed. The general movement was toward improved training in medical school. The third theme was cost. Key informants reported birth defects and genetics conditions services are expensive with public and private insurance failing to cover the charges. Furthermore, costs of services are directly linked to travel distance and under-insurance or total lack of insurance.

4.2 First Steps Early Intervention System Study

The identification of needs associated with genetic deficiencies in the population was enhanced by identification of information already collected by other state entities. The Bureau of Child

Development, Division of Families and Children, Family and Social Service Administration has collected substantial and helpful information in its attempt to develop an evaluation system for its First Steps Early Intervention system. Between February 7, 2000 and June 30, 2000 a total of 17 forums were held across the state focusing on issues of concern for families, children and communities in identifying need, developing services and providing access to community services for disabilities. The Indiana Institute on Disability coordinated these forums that include approximately 300 participants. The focus of these activities was not specifically on genetic illnesses or disabilities, but the types of information collected overlap many of the concerns likely to emerge from families facing the consequences of illnesses associated with genetic origins. The Indiana University Bowen Research Center (IUBRC) team examined reports from this process to determine if further focus groups with consumers/providers scheduled for this project would be duplicative. Examination of available reports indicated that substantial information containing perceptions of the need for and role of community and social services are already available from these forums.

The data from the 17 focus groups identified themes surrounding information needs of families about disease/disabilities and possible responses, availability of support services (mostly social rather than medical), and community responses. The four most common issues for families identified in the 17 forums were:

1. Families knowing about and accessing resources when needed (ranked as one of the top two concerns in 11 of the 17 forums). The commentary surrounding this issue focused on knowledge about resources and how to access them and on actual availability (service is offered).
2. Families advocate by exercising their rights in choosing goals, services and supports (ranked as one of the top nine of 17 forums). Families who are facing the consequences of disabilities and illnesses, many of which are associated with genetic origins, desire accurate and understandable information about these processes in order to appropriately advocate for their child or family member's needs and service regimen.
3. Families express understanding of their child's disability (ranked as one of the top five of the 17 forums). Specifically mentioned were knowledge of the medical condition, prognosis, needs support groups, etc., with emphasis on knowledge about specific diagnoses and long-term implications.
4. Families are connected to other families and natural community supports for emotional support. Families impacted by the disease or disability consequences desire knowledge about networks of support from other families, communities, agencies, schools, etc.

Other family themes focused on maintaining positive and nurturing relationships with affected individuals, ability to maintain normal family activities and routines, ability to participate in community settings and activities, families remaining together, understanding transitions that occur throughout the life of individuals affected with disease/disability, and expectations for satisfactory services.

Other key concerns identified specifically for children focused on goals of independence for individuals with disabling conditions, attaining developmental skills, and participation in community activities. Community concerns focused on the need for communities to welcome and fully include individuals with disabilities and their families through childcare, transportation, retail, housing and employment support. Participants identified the need for communities to provide resources regardless of age, income, insurance status and self-help status.

4.3 Physician Provider Survey

The initial proposal for the genetics project called for a set of focus groups to be performed identifying 30 providers across the state in order to determine the perceptions of service needs and gaps. After completion of the key informant survey and the utilization of the First Steps forums, project staff developed an electronic survey format to reach a wider audience of providers. Membership lists were obtained from the Indiana Medical Society identifying family practitioners and other physician members who would be most likely to encounter initial contact with problems related to genetic deficits. The survey was developed from pertinent research literature and reviewed by geneticists and family practitioners for reliability and face validity. Although previous research has documented the consistently poor response rate common to surveys of physicians, the expected return would still surpass the number of 30 that would have been identified for focus group interviews. Given the electronic format and reliance on e-mail for contact, the data gathered could not be considered representative of the general physician members in the state. However, focus groups also are not generalizable to larger populations. Thus, the decision was made to contact more physicians hoping for a more extensive feedback than would be possible with focus group formats.

In July, the electronic survey format was approved and e-mailed to the mailing list of Indiana family practice physicians. Responses were received from 71 physicians in 29 counties. Ninety percent specialize in family medicine, and almost two-fifths graduated since 1990. Almost three-quarters of the respondents were male. The majority reported their practice panel size as 2501-5000 (39%) or 1001-2500 (26%).

The questions in the survey focused on newborn screening, adult screening, access issues and educational needs. The following were the key findings of the survey:

* Twenty-one percent (21%) of the physicians deal with problems associated with diseases identified through newborn screens at least once per month or more.

* Almost three-fifths have very high or high confidence in the analytical validity of test results from labs, but twenty-one percent (21%) prefer retesting if tests are positive (Figure 4.1).

* Adults with family histories of genetic-related problems are the most likely to request genetic screening and appear to be better informed about the process than other groups. Expectant parents also demonstrate some interest in the process, but over half are unfamiliar with the process (Figure 4.2).

* A substantial majority of physicians report fears of potential problems with privacy/confidentiality of findings from genetic screens. Three-quarters believe the information may interfere with insurability, and three-fifths report fears of employment discrimination for individuals. The potential for other forms of discrimination are also noted by the respondents, but no more than one-third of the physicians focus on these categories (Figure 4.3).

* Few physicians integrate routine genetic testing for adult onset problems linked with genetic problems, but a substantial minority utilize such tests when family history indicates the need or when the patient specifically requests such a screen. Illnesses most frequently screened for genetic bases due to family history or patient requests were HNPCC/FAP (colon cancer); BRCA1 and BRCA2 for breast and ovarian cancer; hemachromatosis and Huntington Disease. Maternal serum screens are routinely performed by more than forty percent (40%) of the respondents (Figure 4.4).

* More than four-fifths indicate they have access to genetic counseling or consulting services. Most often the providers of these services are medical geneticists.

* More than half indicate services are within a 30-minute drive. However, more than one-quarter must drive one to three hours for services.

* Although more than half had made no genetic referrals during the past year, forty-seven percent (47%) had made from 1-5.

* Many physicians are unaware of the length of time their patients must wait for a referral service or appointment. Among those who were familiar with wait time, one-third indicated a wait of one week or less, and slightly more than one-fifth indicated waits of one month or more.

* The most frequently mentioned barrier to accessing health and community services was lack of insurance, 73.2 percent (73.2%), with lack of understanding of importance of the services second with 60.7 percent (60.7%). More than half also identified inability to afford services even when insured as a barrier (See Figure 4.5 for a list of all concerns.)

* When asked to rank the top two concerns out of a lengthier list, the lack of community understanding of importance ranked first, 41.1 percent (41.1%), and the lack of insurance ranked second, 39.3 percent (39.3%). Inadequate availability or number of providers was ranked as third in concern, twenty-five percent (25%) (Figure 4.6).

* The most frequently mentioned information resources or support needed was accurate, accessible web-based information, 84.2 percent (84.2%) and CME short courses at professional meetings, 80.7 percent (80.7%). Physicians also however supported better educational resources to share with families (68.4 percent (68.4%) (See Figure 4.7).

4.4 Common Themes and Conclusions

Common themes permeate the three different primary data bases although the participant characteristics differed from group to group. Although respondents demonstrate a strong faith and confidence in individuals currently providing specialty services, most providers, consumers and physicians believe that additional educational training is necessary to improve other providers' skills and to expand the number of trained providers in the state. Financial support is a key factor in developing a better educated/trained workforce. Insurability and costs of services also appear as concerns both for providers and consumers. Both key informants and physicians view lack of education in the community about the importance of genetic testing and services as a major barrier to developing an adequate support infrastructure. The need for more written materials to provide to families was a key request of physicians along with more continuing education training for practicing physicians and more modules or courses integrated into medical school training. Similar wishes and concerns were voiced by participants in the First Steps forums for better information from physicians to enable families to advocate appropriately for their members.

Some gaps in knowledge among physicians appear to exist concerning specific diagnostic tests related to adult onset problems. Although diagnostic testing to identify the specific diseases is considered a standard procedure, the use of the genetic screen associated with some of these diseases does not appear to be as common. Without better data to support the genetic linkages with adult onset disease and to clarify the environmental interaction with the genetic characteristics, these valuable diagnostic tools may be overlooked in treating disorders.

REFERENCES

Krueger, R.A. Focus Groups: A Practical Guide to Applied Research, Sage Publications, Thousand Oaks, CA. 1994.

FIGURES

Figure 4.1: Confidence in Analytical Validity of Test Results from Labs

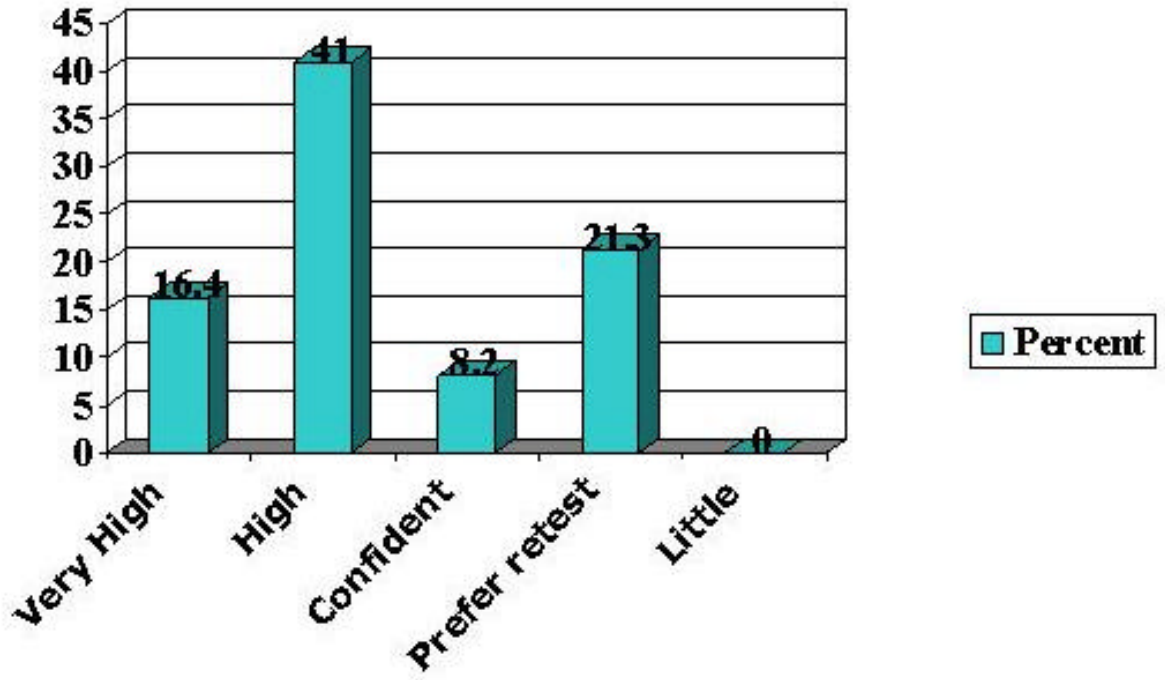


Figure 4.2: Level of interest for genetic screening among patients

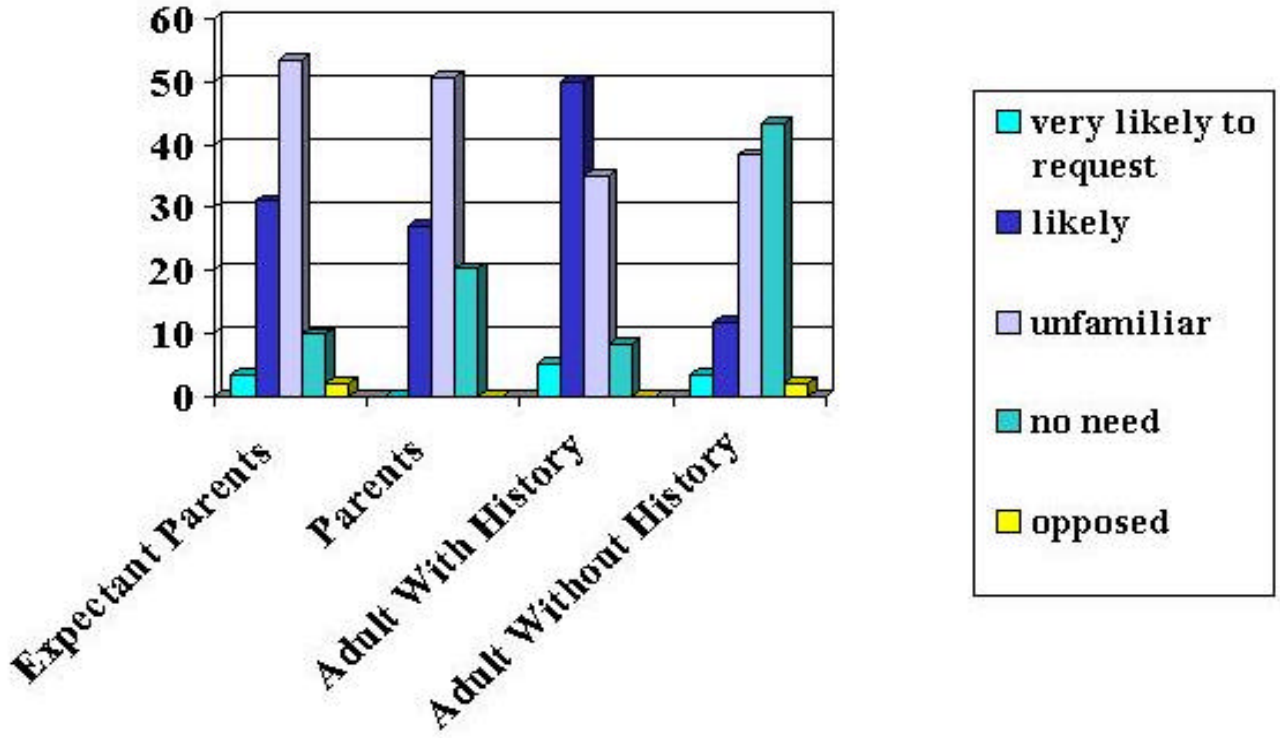


Figure 4.3: Fear Potential Problems with Privacy/Confidentiality

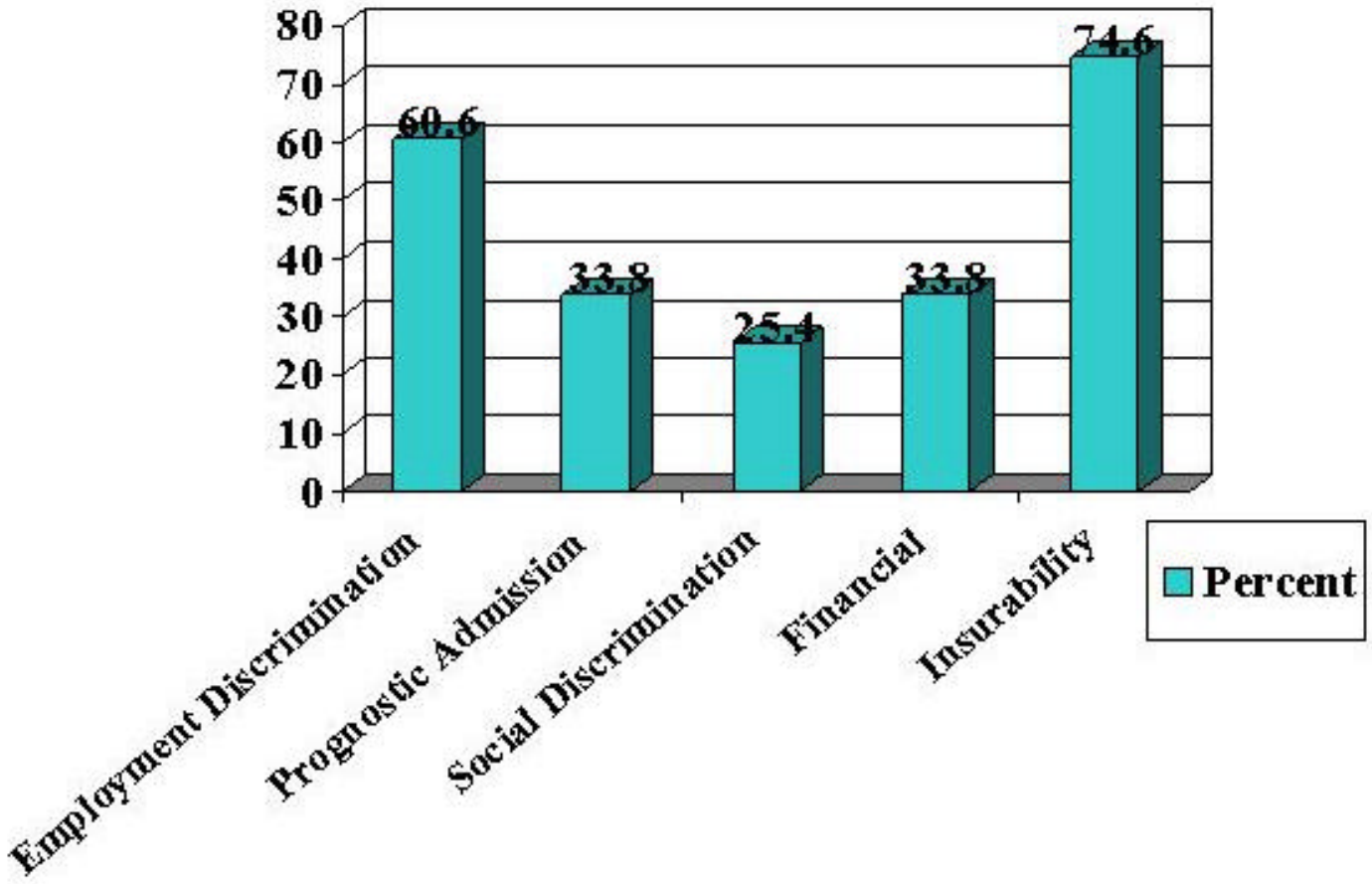


Figure 4.4: Use of Adult Screens in Practice

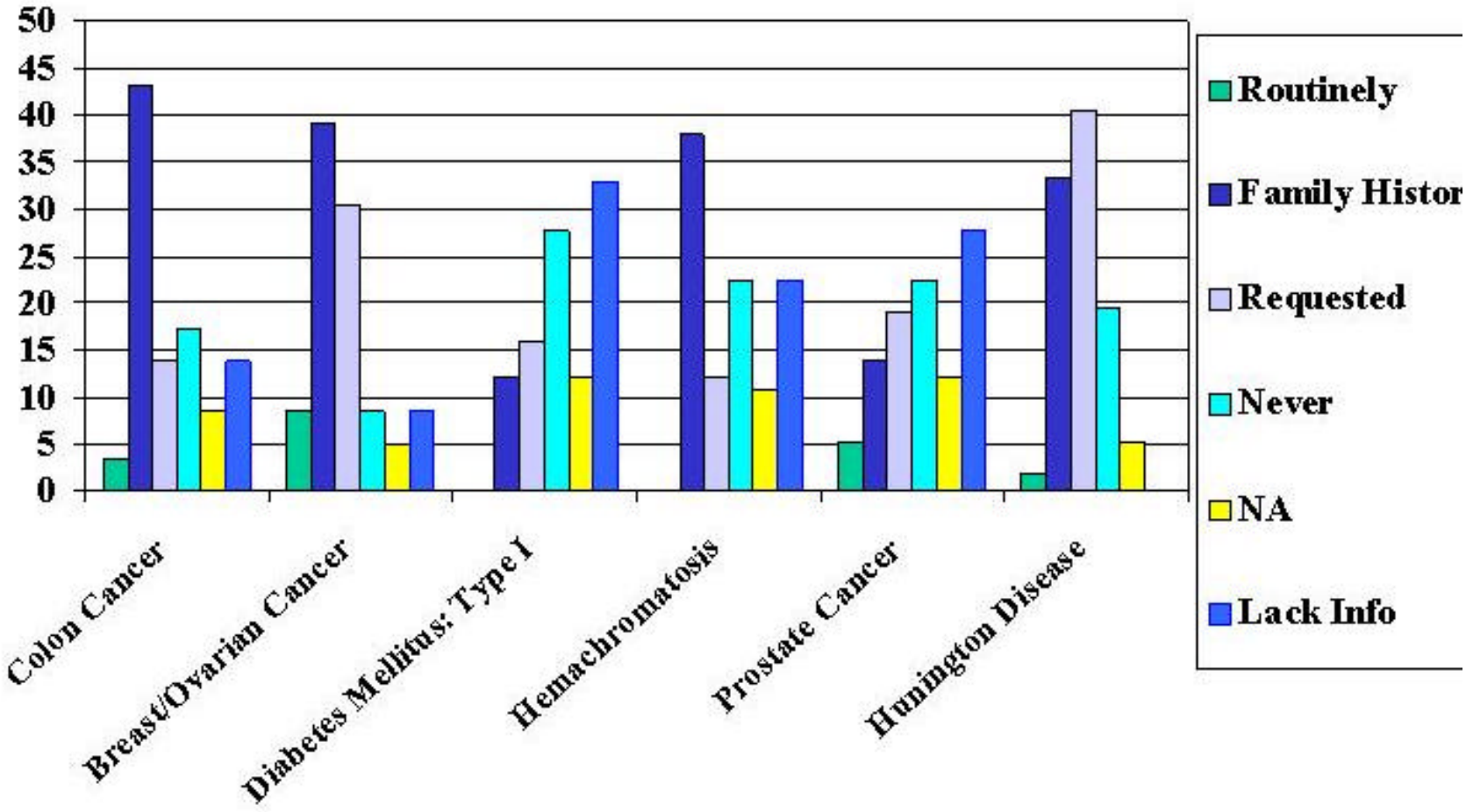


Figure 4.4: Use of Adult Screens in Practice (cont.)

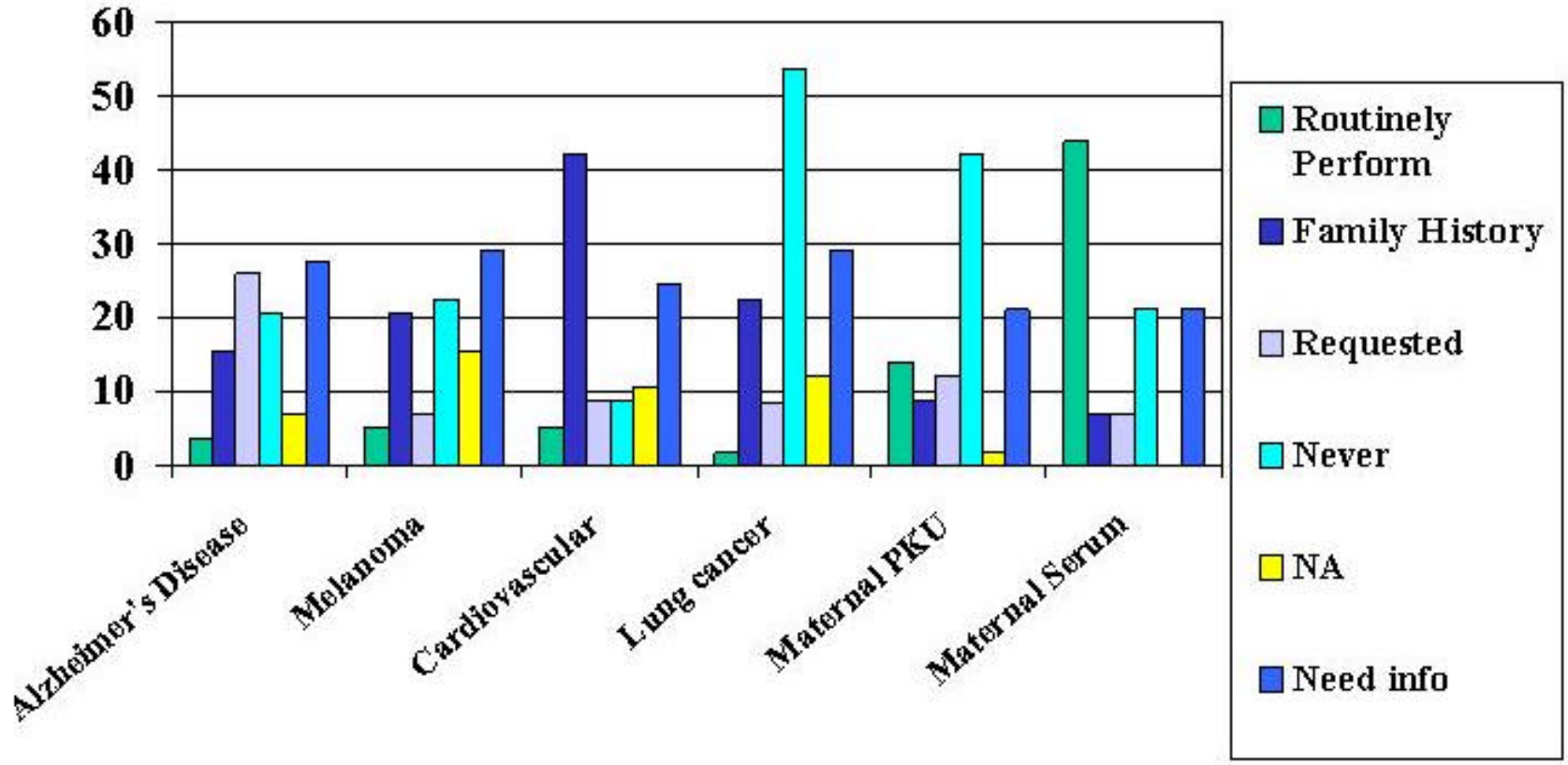


Figure 4.5: Barriers to Health and Community Services

- Lack of insurance, 73.2%
- Lack understanding of importance, 60.7%
- Can't afford even with insurance, 51.8%
- Availability and number of providers, 46.4%
- Inadequately trained providers, 44.6%
- Link to reproductive decisions, 35.7%
- Transportation, 26.8%
- Religious convictions, 25.0%
- Service can't address problem, 14.3%
- Language/cultural barriers, 12.5%
- Other, 8.9%
- Inconvenient times, 5.4%

Figure 4.6: Ranked as One of Top Two Concerns

- Lack of community understanding of importance, 41.1%
- Lack of insurance, 39.3%
- Inadequate availability or number, 25.0%
- Inadequate training, 23.2%
- Inability to pay even with insurance, 21.4%
- Lack faith in ability to address, 12.5%
- Link to reproductive decisions, 12.5%
- Religious convictions, 8.9%
- Other, 7.1%
- Transportation, 3.6%

Figure 4.7: Needed Information Resources/Support

- Accurate, accessible web-based information, 84.2%
- CME short courses at professional meetings, 80.7%
- Educational resources for families, 68.4%
- Valid data bases, 52.6%
- Distance education CME, 43.9%
- Curriculum modules in med school, 29.8%
- Hospital conferences, 29.8%
- Other, 5.3%
- Satellite teleconferencing, 3.5%

5.0 CONCLUSIONS AND RECOMMENDATIONS

The IUBRC team began this project with the goal of applying three different needs assessment models to the data gathering process. A gap method was utilized to identify problems and match service provision patterns to those problems. A marketing approach was utilized through key informant surveys, provider surveys and the use of other forums conducted by state agencies. This concluding section represents the basis for the final perspective, the decision-analysis approach to needs assessment. In this section, patterns and observations from the first two exercises are presented for the use of Genetics Advisory Committee members. All participants in the next phase of the process—that of interpreting, weighing, valuing the evidence and making recommendations –must combine their own experiences to determine what next steps the State of Indiana should take. The IUBRC team will assist committee members in the preparation of final recommendations for a State Genetics Plan that can be used to guide policy decisions for the coming years.

The findings of the gap analysis and marketing assessment identified common themes across several categories: data issues, access issues, educational issues, and privacy/confidentiality concerns. Data concerns can be summarized as follows:

1. The current surveillance systems in Indiana probably result in undercounting of birth defects and genetic disorders. Comparison to active rather than passive systems indicates Indiana's rates to be substantially below other states. Since there are no demographic or population characteristics that would justify this finding, the assumption is that the differences are an artifact of passive versus active reporting and surveillance. IUBRC recommends that the Genetics Advisory Committee examine the possibility of supporting legislation that would require and fund an active system that would improve our understanding of the actual burden of disease and disability in the state. Iowa and Georgia models currently provide the best models available for replication.
2. Data concerning the linkages of adult onset genetic diseases and environmental or behavioral risks are lacking. The Genetics Advisory Committee might explore the possibility of supporting a specialized module on the Behavioral Risk Factor Surveillance System, a data source collected yearly in the state and partially supported by CDC. This module might obtain specific genetic history information for the respondent which could then be linked to behavioral risk activities that are already identified in the survey. Tracked over time, these important data elements would greatly enhance understanding of applied genomics and lead to better service and educational solutions to problems.
3. As the applied genomics field expands with improving technology, an Oversight Committee may be necessary to evaluate the role of public health agencies in performing or supporting tests whose validity and utility must be constantly monitored. The legal, ethical and social implications of producing diagnostic data for problems where no known remedy exists is an ongoing challenge in the field. Indiana experts should play a dominant role in guiding state and local policies and in national debates concerning this important policy issue.

Access concerns expressed by experts, community residents and physicians focused on the following themes.

4. Lack of insurance and inadequate insurance are major barriers to fully accessing the new information technologies that genetic analysis can produce. The Genetics Advisory Committee members must explore strategies to involve the insurance industry as well as public, nonprofit and for-profit providers in supporting better diagnostic data and services.
5. Rural access to diagnostics and service provision is a challenge in Indiana. Prevalence maps indicate that higher rates are not necessarily associated with urban locations, but small numbers make the market reality for provision of services a major challenge. The Genetics Advisory Committee members should explore funding sources for additional outreach services from existing networks to increase access for underserved areas in the state.
6. To assist in capacity building for support services as well as diagnostic and clinical services, the Genetics Advisory Committee members should explore possible partnerships with FSSA First Step Councils. These councils are in place for local planning and ongoing capacity assessment. Participation in these local capacity building networks is an efficient and reasonable approach to enhancing local service provision and addressing access issues. These local councils provide a unique opportunity to explore public and private partnerships at the local level to solve local problems and decrease barriers.

Educational concerns identified in this study focus both on physician training and on community knowledge.

7. Although existing providers are viewed as providing excellent service, the perception remains, both in the community and among physicians, that providers are inadequately trained to meet the needs of individuals with genetic-based diseases. Additional classes in medical school are supported as are the development of CME credit courses linked to professional meetings or CME web-based course that providers can access easily.
8. Community forums and provider surveys both identified lack of understanding in the community about the nature and importance of these problems as a major barrier to provision of appropriate services. The development of educational campaigns that inform the public about new advances and technologies and the importance of risky behavioral activities that interact with genetic predispositions is identified as a major need.
9. Physicians identified better teaching materials for patients as a major need, and this perception was confirmed by participants in community forums who wished for better information about the diagnoses and the long-term impact of disease to assist them in advocating for their family members.

Concerns with confidentially and the adverse impact of diagnostic information on the lives of individuals permeated the different survey groups and forums.

10. As funding and political support is sought for strengthening Indiana's surveillance activities, stringent privacy guidelines must be developed and enforced to protect individuals from adverse consequences in insurability, employability and social participation.

Advisory Committee members are urged to evaluate the various findings from this report, review the similar programs that exist in other states (summarized in Appendix A), weigh the findings and prioritize recommendations as they feel is most appropriate for the state. As the Genetics Advisory Committee groups meet across the next months, the various groups should consider these themes and recommendations and develop policies that will guide the state across the coming years. By February, sub-committees should have a working draft of specific recommendations that the IUBRC group can integrate into a final report for presentation in Spring, 2002.